Citizenship, Genomics, and Mental Health: An Empirical Bioethics Study of Young People's Attitudes towards Advances in Autism Genomics

Arianna Manzini

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Abstract

Background: This empirical bioethics study is motivated by a commitment to involving young people, who are genomic citizens 'in the making', in the bioethics debate around psychiatric genomics. Central to discussions of this kind is the concept of biological citizenship, and its related notion of genetic responsibility, which in Nikolas Rose and Carlos Novas's formulation prescribes biological citizens to actively seek genomic information, and engage in risk calculations to maximize health outcomes and minimize the chances of illness. The focus of the present investigation on psychiatric genomics, and the involvement of children, cast doubts on some assumptions underpinning Rose and Novas's account, which should therefore be put to the test both in relation to *psychiatric settings* and from the perspective of *young people*.

Aims: (1) To conduct an empirical qualitative study on young people's views of autism and moral attitudes towards using genomic information for the prediction, prevention and treatment of autism, and distil the arguments underpinning young people's views and attitudes; (2) to use the results of the qualitative study to inform an understanding of the meaning that young people attribute to the notions of biological citizenship and genetic responsibility in mental health.

Methods: This research used a case study approach focused on autism genomics. Fifty-six young people between 13 and 18 years old from the general population in England participated in 12 focus groups divided into two parts. Part 1 focused on their views of what autism is and is caused by, and whether they thought autism should be conceptualized as a disorder, disability, or difference. Part 2 investigated their moral attitudes towards: (1) prenatal diagnosis and preimplantation genetic diagnosis to prevent autism before birth; (2) genetic tests performed on young children to assess their susceptibility to autism for early intervention purposes; and (3) pharmacological treatments targeting specific risk markers or subcategories of autistic people with the same genotype.

Results: Young people viewed autism as a 'dimensional' condition on a continuum of traits, severity and causes, which informed their 'uncertainty' about whether autism should be

conceptualized as a disorder, a disability, or a difference. This explains why they did not hold clear-cut attitudes towards interventions to predict, prevent and treat autism, but proposed *prima facie* arguments in favour/against. They used the principles of autonomy, beneficence and best interests to argue that the interventions should be made available. However, their endorsement of a dimensional understanding of autism and their uncertainty about how it should be conceptualized motivated a range of concerns around the potential for imposing 'coercive measures'; the risk of encouraging 'stigmatizing' views of autism; the risk of reducing 'diversity' in society; the impact that the interventions may have on 'identity'; and the 'limited clinical validity and utility' of the interventions. Thus, some participants interrogated the meaning of 'being a good parent'.

Discussion: When discussing their moral concerns around advances in autism genomics, participants did not endorse the ethical framework underpinning Rose and Novas's approach. Rather, young people's 'genomic citizenship in the making' prioritizes an ethical framework that promotes citizens' responsibilities for respecting people's identity, accepting and loving diversity, and rejecting stigma, and moves beyond genetic essentialism and determinism. This research makes the case for an expanded notion of biological citizenship to include the ethical framework promoted by young people's genomic citizenship in the making.

Conclusion: This is the first investigation of how children wrestle with ethical considerations in neuropsychiatric genomics. It makes a theoretical contribution to the literature on biological citizenship, by deriving an empirically grounded conception of what being a biological citizen in mental health means to young people. This thesis highlights the considerable potential for the involvement of young people in bioethics research and debates, and lays the groundwork for future opportunities to consider what bioethicists can learn from listening to young people's views of and attitudes towards currently pressing debates.

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List of abbreviations

ABA	Applied Behavioural Analysis
ACMG	American College of Medical Genetics and Genomics
ADHD	Attention deficit hyperactivity disorder
ASD	Autism spectrum disorder
CGT	Clinical genetic testing
CMA	Chromosomal microarray analysis
CNV	Copy number variation
CUREC	Central University Research Ethics Committee
DNA	Deoxyribonucleic acid
DS	Down syndrome
DSM	Diagnostic and Statistical Manual of Mental Disorders
DZ	Dizygotic
EB	Empirical bioethics
EI	Early intervention
EIBI	Early Intensive Behavioural Intervention
EPQ	Extended Project Qualification
FDA	Food and Drug Administration
FGD	Focus group discussion
FX	Fragile X
GCSE	General Certificate of Secondary Education
GWAS	Genome-wide association study
HIV/AIDS	Human immunodeficiency virus infection and acquired immune deficiency syndrome
HR	High risk

IACC	Interagency Autism Coordinating Committee
ICD	International Classification of Diseases
MMR	Measles, mumps, and rubella
MZ	Monozygotic
NBS	Newborn screening
NEUROSEC	Neuroscience, Ethics and Society
NeurOx YPAG	Neuroscience, Ethics and Society Oxford Young People's Advisory Group
NGS	Next-generation sequencing
NHS	National Health System
NICE	National Institute for Health and Care Excellence
NIPT	Non-invasive prenatal testing
NT	Neurotypical
PGD	Preimplantation genetic diagnosis
PGT	Prenatal genetic testing
PND	Prenatal diagnosis
PNS	Prenatal screening
RCT	Randomized controlled trial
SEN	Special Educational Needs
SES	Socioeconomic status
SNP	Single-nucleotide polymorphism
UNCRC	United Nations Convention on the Rights of the Child
VUS	Variants of unknown significance
YPAG	Young People's Advisory Group

Chapter 1. Introduction

This thesis falls under the tradition of empirical bioethics, and adopts a crosscutting approach that spans various academic strands (Figure 1): the literature on biological citizenship; the scholarship on the ethics of neuropsychiatric genomics; and the growing academic interest in young people's participation in currently pressing moral, political and societal debates. The aim of the present chapter is to introduce the different components of this theoretical framework, and explain how they fit together in this empirical bioethics research. This chapter is divided into three sections. Section 1.1 presents the background and rational of the present research; section 1.2 describes the empirical bioethics study that I undertook; and section 1.3 provides an overview of each chapter.

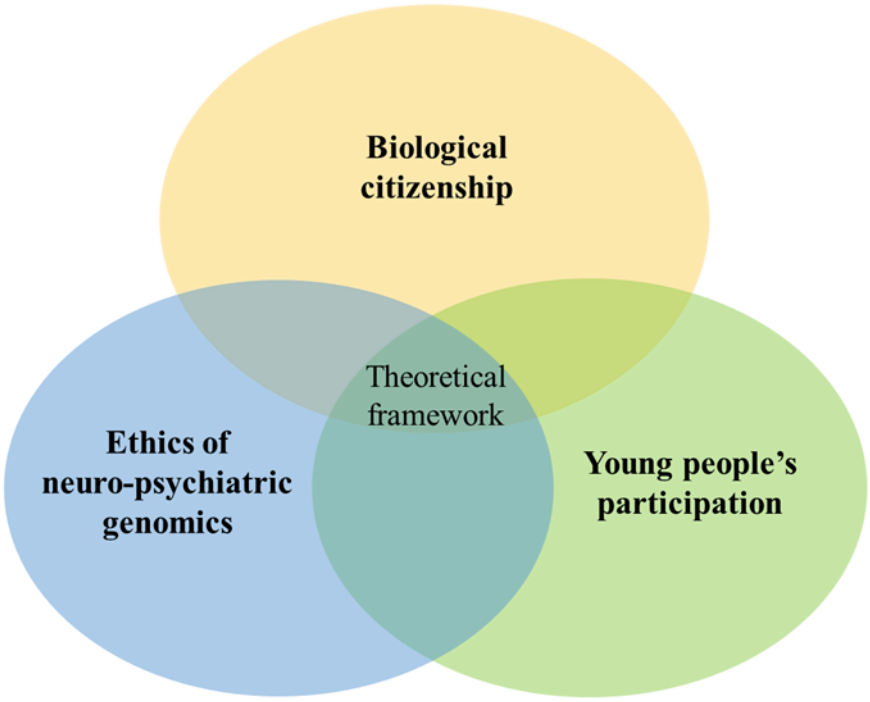


Figure 1: The theoretical framework of this research

1.1. Background and rationale of this research

The present research was motivated by a commitment to involve young people in bioethics debates to understand their values and moral attitudes around the most recent advances and innovations in genomic science, with a specific focus on the intersection between genomics and mental health. Central to discussions of this kind is the concept of biological citizenship, and its linked idea of genetic responsibility. Although many authors have contributed to the literature on this concept, with some scholars referring to it as 'genetic citizenship' (Heath et al., 2008), this thesis focuses on Nikolas Rose and Carlos Novas's overlapping formulations (Rose, 2007; Rose and Novas, 2005), which I will briefly introduce in this section and in the remainder of this thesis I will refer to as the 'traditional notion' of biological citizenship.

By reflecting on the advances made in biomedicine during the 20th and 21st centuries, which other theories of citizenship have neglected, Rose and Novas have argued that biological forms of citizenship link their conception of what it is to be a citizen to 'beliefs about the biological existence of human beings' (2005, p. 440). Although they specify that theirs is a *descriptive* rather than a normative definition of citizenship, the kind of power exercised within biological citizenship is 'intrinsically ethical' (Rose, 2001, p. 42), in the sense that biological notions have become the norm to distinguish between conduct that should be celebrated and conduct that should be blamed, i.e. between morally good and morally reprehensible citizens.

Rose and Novas have focused in particular on advances in genetics to argue that within biological citizenship projects citizens are invested with 'genetic responsibility'. This means that autonomous, self-governing and prudent, yet enterprising, citizens, who use biological language to understand and describe themselves, have a responsibility to actively seek biological (genetic) information about themselves or their family members. Citizens are expected to become informed about their current illness or susceptibility to future ones, and act as rational consumers by maximizing their

chances for health and managing their risk of illness (Novas and Rose, 2000). Because of the normative connotation that biological and genetic notions have acquired in the biomedical age, such responsibility becomes a moral duty or obligation, and so negative judgements are directed towards those who do not live up to the expectation of drawing on genetic information to maximize their health and wellbeing (Rose, 2007).

Rose and Novas's account of biological citizenship has been criticized, and Chapter 2 will provide plenty of opportunities for reviewing scholars' (and my own) criticisms. However, differently from other descriptions of biological citizenship (Heath et al., 2008; Petryna, 2002), Rose and Novas's considerations are not tied to any specific 'biosociality' (Rabinow, 1996), i.e. a group of people who identify with a shared personal or family diagnosis of a certain disease; nor to an ethnographic work situated within a specific geographical area and linked to a specific historical event. Therefore, as has been noticed (Mulligan, 2017), Rose and Novas have made larger claims that apply to any citizen of western democracies in the age of biomedicine. This, I argue, is what makes the traditional notion of biological citizenship relevant to current times.

Indeed, until a few years ago, genetics was almost entirely focused on rare monogenic diseases (e.g. cystic fibrosis or Huntington's disease), which are caused by mutations in a single gene, and so was the ethics debate on genetic research and its implications in clinical practice (the 'gen-ethics debate'). In the early 2000s, improvements in sequencing technologies and advances in study design, particularly the introduction of genome-wide association studies (GWAS), helped scientists progress in their understanding of the genetic contributions to common and complex disorders (Torkamani et al., 2018), i.e. conditions caused by multiple genes (polygenic) as well as environmental and lifestyle factors (multifactorial), and resulting from mechanisms of gene–gene and gene–environment interaction, and epigenetic processes.[1] These advances made it possible to

[1] Although still widely used, the distinction between 'rare single-gene diseases' and 'common and complex disorders' is not as clear-cut as previously thought. Genomic research has recently shown that most disorders are on 'a continuum of common low-risk to rare high-risk genetic variants' (Torkamani et al., 2018, p. 582).

shift from genetics, which studies specific genes and how they are passed down to the next generation, to genomics, which studies the entire human genome or its coding regions. Consequently, new nuances shed light on old gen-ethics concerns (Horn and Parker, 2018; Howard et al., 2015), which prompted ethicists to expand the scope of their research to include genomic practices for common and complex conditions under their normative analysis.[2]

As a result of the shift from genetics to genomics, genomic findings and interventions are (or are predicted to become) relevant to an increasing number of healthcare settings (Davies, 2017),[3] and so to the health and wellbeing of a wider group of stakeholders extending beyond people with a genetic condition, a family history of a certain genetic condition, or professionals invested in the study of genetic conditions only. Some have argued that key stakeholders in genomics now include 'patients, research participants, the public, providers, researchers, advocacy groups, payers, policy makers, and others' (Lemke and Harris-Wai, 2015, p. 949). Potentially anyone, including those who do not have any direct relation to genomic disorders, could encounter genomic information and could be asked to make decisions about it throughout their journey in the healthcare system, which makes Rose and Novas's notions of biological citizenship and genetic responsibility still worth considering. In fact, the shift from the study of rare single-gene conditions to common complex disorders makes it important to reflect on how such an increasingly varied group of people relate to the traditional notion of biological citizenship.

Psychiatric conditions are among the common complex disorders that have recently attracted scientists' as well as bioethicists' interest. In the past two decades, unprecedented research effort has been invested in neuropsychiatric genomics (Corvin and Sullivan, 2016) to align psychiatry to the rest of medicine, in which diagnosis, prognosis and treatment are usually grounded on

[2] In the remainder of this thesis, 'genomics' will be preferred over 'genetics' unless the difference between the two terms needs to be remarked.
[3] For a critical account of this promissory discourse, see for instance Weiner et al., 2017.

knowledge of the aetiological and pathophysiological processes underpinning diseases, rather than on the observation and interpretation of clusters of clinical and behavioural symptoms only.

The initial optimism about the application of genomics to mental health had to inevitably clash with the complexity and difficult interpretation of genomic information in psychiatry. Thus, efforts in psychiatric genomics are now part of broader research programmes that investigate the role that neurobiological contributions more generally play in the development of mental disorders (Appelbaum, 2017). However, ignited by scientists' 'promissory' discourses (Smart and Martin, 2006), the hope remains that the integration of different biological markers for psychiatric conditions could ultimately lead to: (1) more objective diagnosis, by moving from a classification of psychiatric disorders based on behaviours to one based on laboratory measures (Haag, 2006); (2) early and accurate prediction of susceptibility to mental illness or prognosis grounded on biological indicators, to allow prevention and early intervention (Hyman, 2003; TedBlog, 2013); and (3) more effective psychological and pharmacological treatments that target subcategories of patients sharing similar genotypes or underlying biology (Insel, 2014).

Ethicists' close scrutiny of this new branch of genomics was not long coming, as shown by several papers being published since the early 2000s (see Appelbaum, 2004; Kong et al., 2017; Kong and Singh, 2018; Manzini and Vears, 2018; Singh and Rose, 2009) and research projects being developed on the ethics of neuropsychiatric genomics.[4] Many empirical studies have also been conducted to investigate adults' attitudes towards translational applications of psychiatric genomics research (see Laegsgaard et al., 2009; Wilde et al., 2010). However, no systematic research has so far been conducted to understand young people's moral concerns in the context of the recent

[4] See for instance the *Becoming Good: Early Intervention and Moral Development in Child Psychiatry* project (www.begoodeie.com), of which the present research is part; the *Center for Research on the Ethical, Legal and Social Implications of Psychiatric, Neurologic and Behavioral Genetics* at Columbia University (http://braingenethics.cumc.columbia.edu/); and the *Global Initiative in Neuropsychiatric GenEthics* a collaboration among the University of Oxford, the Stanley Center at the Broad Institute of Harvard and MIT, and local mental health researchers (https://neurogene.com).

advances in psychiatric genomics, and what these tell us about the meaning that young people attribute to biological citizenship in psychiatric contexts.

As I will discuss in Chapter 2, social scientists' recent interest in young people's participation has revealed that children's contributions have for long been overlooked in social, political and moral debates due to assumptions about their vulnerability and incompetence, which translate into recurrent scepticism over their capacity to reflect on complex issues (Singh, 2016a; Singh and Keenan, 2010). However, I argue that the exclusion of young people, among other biological citizens, from ethics debates about neuropsychiatric genomics is particularly problematic because children are 'genomic citizens in the making' (Sabatello and Appelbaum, 2016). This means that they constitute 'the generation most likely to be affected' by interventions made available by our increasing knowledge in genomic science (Iredale et al., 2010, p. 49), including recent advances in psychiatric genomics. As a consequence of these advances options that were once unavailable in psychiatric settings have become possible or are likely to become so, which will require today's children to deal with ethical questions that were less (or not) relevant to older generations. Thus, it is important to create the space for young people to express their views and attitudes, with a specific focus on what they value as normatively important in discussions around psychiatric genomics.

Therefore, this thesis uses autism genomics as a case study to investigate the ethical concerns that young people, i.e. genomic citizens in the making, express in the context of the application of genomic science to psychiatric conditions. In this way, this thesis puts Rose and Novas's traditional notion to the test, by considering what young people's moral concerns and ethical arguments tell about the meaning that the current generation of young people attribute to biological citizenship and genetic responsibility in mental health settings.

1.2. Description of this research

This is an empirical bioethics study that focuses on the practical ethical context at the intersection between genomic science and mental health, which is investigated through the lenses of the scholarship on biological citizenship (Figure 2). This is the first research to put Rose and Novas's account of biological citizenship to the test both in relation to psychiatric settings and from the perspective of young people. In what follows, I provide a rationale for choosing autism as a case study among other psychiatric conditions, which will help to clarify the aims, objectives and research questions of this study. I then discuss the contribution that an empirical bioethics study provides to this area of bioethical inquiry, and I justify why this empirical study focuses on the general population of young people. Because there will be many opportunities to discuss these theoretical and methodological choices in depth in the next chapters, here they are only briefly presented to provide an overview of the present research and set out the main conceptual commitments underpinning this thesis.

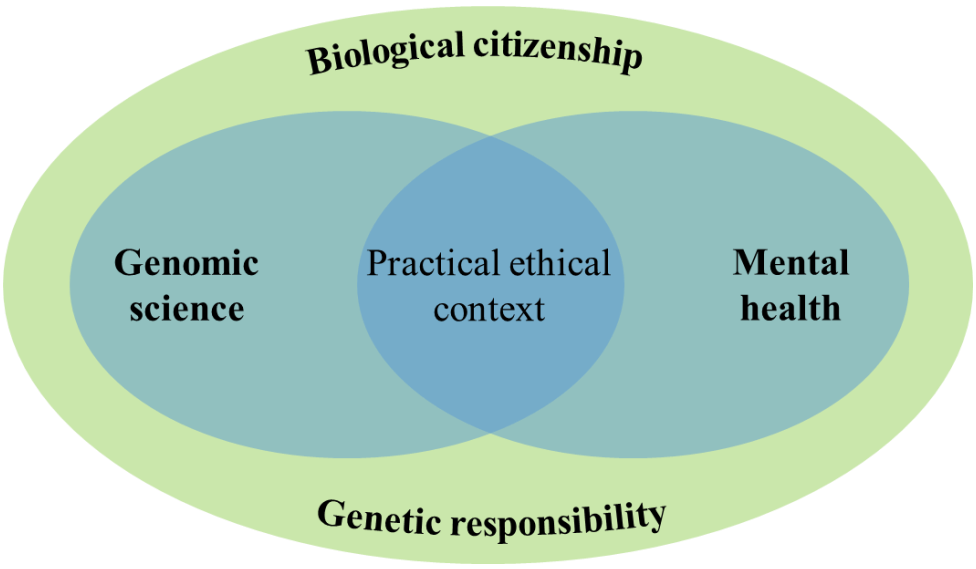


Figure 2: The practical ethical context at the core of this research

1.2.1. Autism genomics as a case study

In western medicine, what we currently call autism spectrum disorder (ASD) started to be investigated in the 1940s, when psychiatrist Leo Kanner brought autistic traits to the attention of the medical community (Kanner, 1943). According to the latest edition of the *Diagnostic and Statistical Manual of Mental Disorders* or DSM-5, autism is a neurodevelopmental disorder whose heterogeneous symptoms manifest across a spectrum of severity. However, the autism diagnosis includes two core features, i.e. 'persistent deficits in social communication and social interaction across multiple contexts' and 'restricted and repetitive patterns of behaviour, interests, or activities' (American Psychiatric Association, 2013, p. 50).

Within a discussion of the ethics of neuropsychiatric genomics, ASD offers a good case study because, since twin studies in 1970s, it has been the focus of increasing interest of genetic and genomic research aimed at developing targeted pharmacological treatments, preventative measures and early interventions (Singh, 2016b). This has caused a range of ethical concerns, primarily due to the enduring profound disagreement on how autism should be conceptualized despite widespread public and scientific interest in the topic. As Nadesan acknowledged: 'Even at the dawn of the twenty-first century, we do not know what autism *is*' (2005, p. 9).

Twenty years into the new century, it could be argued that not much has changed since those words were published. Autism is still conceptualized as a disorder to be predicted, prevented and treated under the medical model; or a disability to be accommodated under the social model; or a difference to be celebrated under the neurodiversity model. The lack of consensus around the nature of ASD among different stakeholders means that the question of how (and in fact whether) autism should be treated, prevented, or intervened in early in life, has fuelled conflicts between opposing views. Among these, a strongly critical perspective against the increasing interest of genomics in autism has been raised by autistic adults' self-advocacy organizations, who understand it as an expression of a societal desire to eliminate autism due to mistaken assumptions about the value of autistic lives

(Orsini, 2009; Ortega and Choudhury, 2011). In this sense, autism is one of the best examples of those psychiatric conditions that puzzle and fascinate scholars working on the ethical and social dimensions of neuropsychiatric genomics.

Given the complexity that characterizes public discourses around autism, in the autism literature language is considered particularly sensitive, because different terms are thought of as expressing different values about and attitudes towards autism, and so some terminological clarifications are essential at the outset of this thesis (see Table 1). If in some passages the medical language is used as a shorthand, this should not be interpreted as casualness towards the complex naming and identity politics that characterizes autism.

Table 1: Glossary of autism-related terms used in this thesis

Term	**Description**
Autism, autism spectrum disorder, ASD	These terms are used interchangeably unless otherwise specified
People with autism, autistic people, autistics	These terms are used interchangeably unless otherwise specified, although I acknowledge that some within the autism community prefer the person-first language (i.e. the first term), while others the identity-first language (i.e. the second and third terms) to indicate that autism is an integral part of the individual, rather than an appendage to it (Walsh, 2010)
Neurotypicals, NT	This term and acronym are used to refer to individuals who do not display autistic patterns of behaviour
Psychiatric condition, mental condition	These terms are used as the most generic and value-free phrases to refer to autism without attaching any normative connotation to it
Disorder, illness, psychiatric/mental disorder, psychiatric/mental illness	These terms refer to psychiatric conditions under the medical model, or in other words to mental traits that are considered inconvenient and warrant medical attention
Mental disability	This term is used to refer to the social model, which sees mental difficulties as the result of societal barriers

1.2.2. Aims, objectives and research questions

This research investigates young people's moral attitudes towards interventions that propose to use the knowledge of autism genomics to prevent or to treat the condition, or to intervene early to lessen autism risk.[5] Because societal desire for these interventions is likely to be influenced by the way in which autism is conceptualized, a preliminary investigation of whether young people view autism as a disorder, a disability, or a difference is deemed necessary. Thus, this research provides a picture of young people's views of autism and attitudes towards the interventions mentioned above, and it particularly focuses on the arguments underpinning such views and attitudes. By situating the research findings within the literature on biological citizenship, particularly Rose and Novas's formulation, the aim of this thesis is to consider what young people's arguments tell about the meaning that the current generation of young people attributes to biological citizenship and genetic responsibility in mental health settings. Finally, because this is an empirical bioethics research, by integrating the empirical data with philosophical and ethical analysis, this thesis draws normative conclusions about why and how the responsibilities promoted by young people should underpin and guide action in psychiatric genomics settings. Therefore, the primary research questions of the present research are as follows:

1. What meaning does the current generation of young people attribute to 'biological citizenship' in mental health settings?
2. What responsibilities do contemporary young genomic citizens promote under this account of biological citizenship?

[5] Please note that in this thesis the following expressions will be used as synonyms: interventions that propose to use the knowledge of autism genomics to prevent or to treat the condition, or to intervene early to lessen autism risk; interventions that propose to use genomic information to prevent autism, treat it, or predict it to lessen its risk; interventions for the prediction, prevention and treatment of autism; interventions for the prevention of autism at different life stages; interventions for the primary and secondary prevention of autism.

3. Why and how should these responsibilities underpin and guide action about the use of genomic information in psychiatric settings?

Table 2 provides a detailed account of the research questions, aims and objectives of this thesis.

Table 2: Research questions, aims and objectives of the present research	
Primary research questions	• What meaning does the current generation of young people attribute to 'biological citizenship' in mental health settings? • What responsibilities do contemporary young genomic citizens promote under this account of biological citizenship? • Why and how should these responsibilities underpin and guide action about the use of genomic information in psychiatric settings?
Secondary research questions	• How do young people view autism? • What moral attitudes do young people hold towards interventions that propose to use genomic information to prevent autism, treat it, or predict it to lessen its risk?
Aims	To investigate the meaning that young people attribute to the notions of biological citizenship and genetic responsibility in mental health, and draw normative conclusions on why and how their contributions should underpin and guide action about the use of genomic information in psychiatric contexts.
Objectives	• To conduct a review of the literatures on biological citizenship, the ethics of neuropsychiatric genomics and children's participation; • To conduct a review of the ethics, sociological and scientific literature on autism, and so provide a justification for the choice of autism genomics as a case study; • To conduct an empirical qualitative study that generates a picture of young people's views of autism and moral attitudes towards using genomic information for the prediction, prevention and treatment of autism; • To conduct a qualitative analysis and philosophical analysis of the empirical data to distil the arguments underpinning young people's views and attitudes and identify where and why young people might have promoted invalid or unsound arguments; • To use the results of the analysis to investigate the meaning that 'biological citizenship' and 'genetic responsibility' have to the current generation of young people in mental

	health, and draw normative conclusions on why and how young people's contributions should underpin and guide action about the use of genomic information in psychiatric contexts.

1.2.3. An empirical bioethics study that focuses on the general population

This thesis does not aim to perform 'hard' normative work, whereby empirical data are used to develop a decisive conclusion or strong argument on the 'right' way to conceptualize autism and the 'right' choices that biological citizens should make in the context of autism genomics. Rather, the aim of the philosophical analysis performed in this research is to elucidate the moral concerns that young people raise about advances in autism genomics, to inform an understanding of the way in which they think of biological citizenship in mental health contexts, and thus discuss why and how their reflections should be used to guide choices in the practical ethical context at stake. To this purpose, young people's empirically derived accounts are put under philosophical scrutiny to identify where and why participants might have promoted arguments that fail to meet the universal standards of reasoning in moral argumentation (i.e. validity and soundness).

In this research, moving beyond an investigation of young people's views and attitudes to conduct a normative analysis of the empirical data is necessary for two main reasons. First, examples of harmful practices that were used in the past to 'treat' autism, and are still used in some places (e.g. packing therapy, see Spinney, 2007), are an important reminder that scrutiny of society's moral values is fundamental in psychiatry. If certain practices were once considered acceptable, so may be other harmful interventions made possible by genomics. Second, as will be discussed in this thesis, a recurrent assumption in public and academic discourses is that children do not have the capacities to contribute to discussions about complex ethical issues in a meaningful way. Therefore, identifying where young people's arguments are (or are not) valid and sound is important to

anticipate and address potential questions about why their contributions should be taken seriously to draw conclusions in this thesis.

It is important to note that participants in this study are young people from the general population. A review of the sociological and ethics literature on this topic (see s3.5) revealed that, at the time when this research started in September 2017,[6] much ethics research on psychiatric genomics had focused on autism and the implications of advances in autism genomics, but hopes, expectations and concerns in relation to such advances had been generally discussed with autistic people, their family members and professionals invested in the study of autism (see Hens et al., 2016b; Li et al., 2016; Pellicano et al., 2014). Less attention had been paid to the views and attitudes held by the general population, by which I mean people who have no special relation to autism by virtue of being on the autism spectrum, having a family member with autism, or being invested in the study of autism (the 'experts', such as academics, psychiatrists, geneticists, etc.).[7]

This gap cannot just be accounted for by the assumption that the general public have little interest in discussions about ASD and ASD genomics. As mentioned above (s1.1), genomic advances are becoming relevant to a wider group of biological citizens, who therefore should be given the opportunity to shape policies and the broader cultural debate about genomic interventions. This is particularly important in psychiatry, which is fundamentally positioned at the intersection between medicine, biological sciences, morality and society (Singh and Wessely, 2015).[8] Because in psychiatry medico-scientific considerations are intertwined with moral, societal and cultural values,

[6] Although I started my DPhil in October 2016, at the beginning of my second year I changed the focus of my research from psychosis to autism, because the rapidly evolving field of autism genomics meant that there was more tangible evidence of advances in autism genomics on which to base an ethical analysis of interventions for the prediction, prevention and treatment of autism; the focus on psychosis would have instead been more speculative. Therefore, the present research started in late September 2017.

[7] Although the notion of a 'general population' as opposed to 'experts' has been contested (see for instance Kerr et al., 1998), in this thesis I will use it as a shorthand to refer to (young) people who have no direct relation to autism, mental health and/or genetic conditions.

[8] Please note that I am not arguing that psychiatry is unique in this respect. As I will discuss in Chapter 2, normative considerations and socio-culturally determined judgments also characterize other fields of medicine. What I am claiming is that this argument definitely applies to psychiatric settings.

the discussion about what behavioural traits should count as 'disorders', whether certain behaviours are normal or abnormal, and in fact whether 'normal' behaviour exists at all, should not involve 'experts' only. On the contrary, it is an endeavour in which everyone should be invested. This suggests that we should investigate what biological citizens, as broadly understood as possible, think of autism and its causes, what value judgements they attach to autism and how they think these judgements should guide the use of genomic information for the prediction, prevention and treatment of the condition. After all, technological advances do not only pose problematic choices and dilemmas to people directly affected by them; they instead have 'a broader cultural life beyond the material – functioning as representations and points of cultural resonance for particular social groups and actors' (Kerr, 2009, p. 62).

This is even more important because autistic people do not live in a vacuum, but in societies with neurotypicals, including the general population that has no specific relation to autism. In fact, some have argued that, because of its behavioural nature, autism would not even be 'a thing' if autistics did not live in societies with neurotypicals – as Starr put it, 'Robinson Crusoe couldn't have been autistic until he met Man Friday' (quoted in Bovell, 2015, p. 80). The same could apply to any condition that affects behaviour, but it may be particularly the case in autism because of the social communication and social interaction difficulties that autistics specifically experience (Davidson and Orsini, 2013), which inherently have to do with the way they socialize with and relate to other people. In turn, this is likely to impact on societal attitudes towards autism.

Therefore, without wanting to neglect the importance of listening to individuals with first-hand experience (e.g. autistic people, family members and autism professionals), in the context of autism it is fundamental to investigate the perspectives that neurotypical 'lay' people, among whom autistic individuals live, hold towards the development of genomic interventions to predict, prevent and treat this condition. This once again confirms the relevance of Rose and Novas's account of biological citizenship as applying to potentially any members of western democracies in the

biomedical age, as well as the significance of investigating what biological citizenship in psychiatric settings means to this group of stakeholders.

A 'methodological' and a 'political' rationales should be added to these considerations, in order to justify the involvement of young people, among other biological citizens, in ethics discussions about neuropsychiatric genomics. From a *methodological* point of view, while systematic research has investigated adults' attitudes, children's voices are still missing from the debate around recent advances in the understanding of the genomics of psychiatric conditions (see s1.1). From a *political* point of view, it is important to create the opportunities for young people to express their moral concerns, because advances in psychiatric genomics are likely to generate changes in psychiatric care and practices that will affect today's children, when they reach the age of majority or decide to reproduce.

In Chapter 2 (s2.4.1), I will explain that this political rationale is part of a broader and increasing academic interest in children's participation in debates about pressing societal and political issues, so to ensure young people's greater role in contributing to policies that may affect them, and to cultural changes that will enable the future they want to live in (Davis and Hill, 2006). Youth activism is already gaining momentum in areas such as education, climate change, gun violence and, more recently, mental health (Singh et al., 2020; The Lancet, 2019). It is within this tradition that I situate the commitment of the present research to understand what young people consider as normatively relevant in psychiatric genomics, so that they can contribute to shape the ethics agenda of practices at the intersection between genomics and mental health.

Having discussed the background, rationale, and aim of the present research, in the remainder of this chapter I provide the outline of this thesis.

1.3. Overview of this thesis

Chapter 2 situates the present research within the literature on biological citizenship. In the first part, this scholarship is critically reviewed, with a particular focus on Rose and Novas's account. Building on criticisms that others have raised, I argue that although still relevant to the current times, the traditional notion of biological citizenship constructs citizens' responsibilities and obligations around three assumptions that need further investigation. These are: (1) an alleged consensus over the distinction between health and illness; (2) a linear relationship between genetic risk and action; and (3) a conformity among duties, choices and values.

By illustrating the various layers of complexity that characterize the field of psychiatric genomics, I question whether the first two points can still be considered defensible given the most recent advances in genomics. By discussing the concept of 'genomic citizens in the making' (Sabatello and Appelbaum, 2016), I question whether the third assumption can account for the way in which young people relate to biological understandings of citizenship. I then pull the various strands of this chapter together to argue in favour of the significance of putting the traditional notion to the test, to assess both whether it fits mental health settings and whether young people discuss psychiatric genomics within the framework proposed by Rose and Novas.

Chapter 3 expands on the rationale for choosing autism genomics as a case study. In the first part I demonstrate that autism fits the theoretical framework developed in Chapters 1 and 2, because of its contested diagnosis, aetiology and nature, and the stigma attached to it. The second part reviews the empirical literature on stakeholders' moral concerns about advances in autism genomics. Three main gaps are identified: (1) most studies have failed to reflect on the value judgements about autism that underpin societal demand (or lack thereof) for interventions to predict, prevent and treat autism; (2) the general population's attitudes towards autism genomics, and young people's

perspectives in particular, have not yet been investigated; and (3) only one study has so far discussed stakeholders' attitudes within the tradition of biological citizenship.

Chapter 4 provides a detailed account of the methodology and methods of the empirical study that I conducted to fill in these gaps, which was developed in consultation with the Neuroscience, Ethics and Society Oxford Young People's Advisory Group. I explain that 13–18-year-old young people participated in focus groups that investigated their views on what autism is, and is caused by, and whether autism should be seen as a disorder, a disability, or a difference; and their moral attitudes towards using genomic information to predict, prevent and treat autism. Three sets of interventions were discussed: (1) prenatal diagnosis and preimplantation genetic diagnosis to prevent autism before birth; (2) genetic tests performed on young children to assess their susceptibility to autism for early intervention purposes; and (3) pharmacological treatments targeting specific risk markers or subcategories of autistic people with the same genotype.

Chapter 5 presents young people's views of autism in light of the two main categories that data analysis identified, i.e. 'dimensionality' and 'uncertainty'. Young people seemed to conceptualize autism as a dimensional condition on a continuum of heterogeneous positive and negative traits; on a spectrum from mild to severe forms; and as characterized by a multifactorial aetiology aligned on a dimension of genetic to environmental factors. Young people's dimensional understanding of autism informed their uncertainty about whether autism should be categorized as a disorder, a disability, or a difference. Within such uncertainty, some participants expressed resistance towards considering autism as a disorder or disability because of concerns about stigmatization.

Young people's dimensional understanding of autism and uncertainty about its nature informed their attitudes towards the three interventions to predict, prevent and treat autism, which are presented in **Chapter 6**. Young people did not hold clear-cut attitudes towards the interventions, but proposed *prima facie* arguments in favour or against. They used the principles of autonomy, beneficence and best interests to argue that the interventions should be made available. However,

their endorsement of a dimensional understanding of autism and their uncertainty about its nature motivated a range of concerns towards using genomic information to prevent autism at different life stages, which revolved around the following categories: 'imposing coercive measures', 'increasing stigma attached to autism', 'impacting on (autistic) identity', 'reducing societal diversity', 'limited clinical utility and validity' and 'being a good parent'.

Chapter 7 interprets the main findings of the empirical study in light of the literatures discussed in the background of this thesis, hence providing an empirically grounded account of what biological citizenship and genetic responsibility mean to young people in mental health. I show that participants' discussions confirm the suspicion that Rose and Novas's traditional notion of biological citizenship is of difficult application to the context at the intersection between genomics and mental health, and does not account for the way in which genomic citizens in the making understand citizens' responsibilities in the biomedical age.

Participants did not endorse the ethical framework underpinning Rose and Novas's account. Rather, young people's 'genomic citizenship in the making' produces a range of responsibilities that rotate around the values of respecting people's identity, accepting and loving diversity, and rejecting stigma; and it moves beyond genetic essentialism and determinism to acknowledge the limited clinical validity and utility of genomic information in psychiatry. Because this framework derives from young people's understanding of autism as a dimensional condition, and their uncertainty about the value judgements that should be attached to it, I suggest that it could be applied beyond mental health to guide action in the use of genomic information about any condition characterized by dimensionality and uncertainty. Thus, this thesis makes the case for an expanded notion of biological citizenship to include the ethical framework promoted by young people's genomic citizenship in the making. This chapter concludes by discussing the limitations of the present study to make recommendations for future research, before **Chapter 8** summarizes the main arguments of this thesis.

Chapter 2. Biological citizenship: Relevance and limitations of the traditional notion

2.1. Introduction

Chapter 1 introduced the theoretical framework underpinning this research, which is informed by different bodies of academic thought. This includes the literature on biological citizenship, which is important to review at the outset of this thesis, as it informs the reflections that will follow.

The concept of biological citizenship appeared in sociological and anthropological writings at the end of the twentieth century, when scholars started to theorize the rise of new ways of thinking of, and exercising, citizenship as a result of the progress made in the understanding of human biology. The concept has been attributed different connotations, but it usually refers to forms of belonging and shared identity that groups of people construct around their common injury, genetic status, or disease to make right claims, and demand social recognition and access to services (Mulligan, 2017; Singh, 2016b). This is how Petryna used 'biological citizenship' in one of the first ever formulations of the concept (2002), to illustrate how people affected by the Chernobyl nuclear disaster demanded medical and social services from the Ukrainian government in the name of their injured (biological) bodies. As a variation of biological citizenship, 'genetic citizenship' was used by authors such as Heath et al. (2008), whose ethnographic work was primarily concerned with the activism of people with rare single-gene conditions and their 'allies', including relatives, scientists, researchers and political representatives.

These are only two examples of a broader range of theories that from the early 2000s reflected on the impact that biological notions have had in shaping ideas of being a citizen. The present thesis focuses on Rose and Novas's contribution to this literature (Rose, 2007; Rose and Novas, 2005), which I will refer to as the 'traditional notion' of biological citizenship. The relevance of Rose and

Novas's account to the present research is because of the larger claims that they made about biological citizens in general (s1.1), by virtue of which their theory fits well with an investigation focusing on the general population. It is also important to note that most bioethics literature engaging with the concept of biological citizenship does so with reference to their writings. In section 2.2, I will critically review Rose and Novas's account in order to reveal its limitations, which, as I will argue, warrant further investigation. These limitations will be illustrated in the context of mental health in section 2.3, and with reference to young people in section 2.4, before this chapter concludes by remarking on the importance of putting the traditional notion of biological citizenship to the test.

2.2. Biological citizenship according to Rose and Novas

Like other scholarly reflections on biological citizenship, Rose's and Novas's writings were influenced by Michel Foucault's theory of *biopolitics* (Foucault, 1979). According to Foucault, modern western states do not ground power on traditional methods of control through punishment and revenge, but impose discipline through practices of 'normalization', i.e. by defining what normal and abnormal conduct is in various dimensions of social and private life. A paradigmatic form of power is *biopower*, which uses scientific knowledge and expertise to impose standards and norms about people's health and wellbeing (Foucault, 1979).

Following Foucault, Rose has argued that modern liberal democracies have developed a way of governing individuals 'at a distance', i.e. by shaping people's attitudes so that these align with the state's political and economic interests in health. In this way, being inculcated into the fears and hopes of citizens, '[b]y the second half of the twentieth century, health had become one of the key ethical values' (Rose, 2007, p. 22). The moral duty to be healthy has grown into an increasing sense of individualized responsibility to take care of one's own health and that of one's family members, which progressively took prevalence over more traditional forms of (mainly) passive obedience to

state-imposed programmes (e.g. eugenic practices). In fact, as Rose has put it: 'The huge extension of the political apparatus of health […] would have been inconceivable had the values of health not become entangled with the aspirations of "the people" themselves' (Rose, 2001, p. 39).

Such an extension consists, in part, in the significant progress made in the life sciences and biomedicine, which has led to a better understanding of the molecular constituents of human life and hence, according to Novas and Rose (2000), to new kinds of relations that people develop with themselves. Not only do biomedical practices envision life at the molecular level, the age of molecular biomedicine has also seen the birth of the 'somatic individual' (Novas and Rose, 2000), i.e. a mutation in personhood whereby people understand their and others' identity or subjectivity in terms of its relation to their body and corporeality. Thus, people increasingly use the language of biology to describe themselves and others, their traits and identities; and refer to the good or ill functioning of the body as the *locus* that drives deliberations about how they could or should act.

Rose and Novas's writings suggest that the combination between the internalization of the state's duty to be healthy and the application of the 'molecular gaze' (Rose, 2007) onto one's selfhood has had implications on individuals' understanding of themselves as members of their social and political communities. It is within this framework that Rose and Novas's argument about the rise of biological citizenship should be interpreted.

2.2.1. Biological citizenship, genetic responsibility, and ethopolitics

Rose and Novas have discussed several reasons for why the conception of citizenship as defined by national borders, which was until recently predominant, has progressively faded away. Among these, they have argued that developments in biology, bioscience and biomedicine, and the increasingly detailed information they produce about citizens' biological life, have played a critical role in the rise of a new kind of social and political agent, which they call the biological citizen. 'Biological citizenship' is defined as including:

> *[A]ll those citizenship projects that have linked their conceptions of citizens to beliefs about the biological existence of human beings, as individuals, as families and lineages, as communities, as population and races, and as a species.*
>
> (Rose and Novas, 2005, p. 440)

Rose and Novas have specified that theirs is a *descriptive* rather than normative definition of citizenship. However, because their reflections are grounded on Foucault's theory of the way in which modern western states impose discipline by instilling norms of conduct in their individual citizens, the kind of power exercised within biological citizenship inevitably has normative implications (Plows and Boddington, 2006). Within biological forms of citizenship, biological notions become the norm (in a Foucauldian sense) to distinguish between 'actual, potential, troublesome and impossible citizens' (Rose and Novas, 2005, p. 440), and so between conduct that should be celebrated and conduct that should be censured, or between morally good and morally reprehensible citizens.

Rose has interpreted these norms in light of the neoliberal frameworks of western democracies, in which biological citizenship has flourished. Such frameworks are characterized by a pervading market rationality, which promotes the maximization of free trade and competition while shifting away from welfarism, and so transfers responsibility (or rational action) for citizens' thriving from the state to its individual members (Brown, 2003). These are required to be citizen-consumers who exercise their calculating skills to assess the costs, benefits and consequences of the options that are available to them, hence taking an active role in the process of self-improvement and contributing to the flourishing of society (Rose, 2018).

Neoliberal rationality translates into an ethical framework typical of biological forms of citizenship that Rose calls 'ethopolitics' (2007). This is an ethics that promotes *active* citizenship in the sense that self-governing and prudent, yet enterprising, citizens have a responsibility to actively seek biological information about their current illnesses and risk of future ones, and behave as rational consumers who take action so to maximize their health outcomes. Because biological information

has individual and familial relevance, biological citizens' responsibility extends to others, particularly their family members. This means that in light of the knowledge of their own susceptibilities to ill health in the future, good citizens are supposed to modulate their life strategies and decisions about jobs, marriage and reproduction.

Biological citizenship is, therefore, *individualizing* because the state's duty to be healthy becomes internalized in such a way that moral values rotate around the individual body, which becomes the *locus* for amelioration of the self. Yet, Novas and Rose borrow Rabinow's notion of 'biosociality' (1996) to remark that biological forms of citizenship are simultaneously *collectivizing* (Rose and Novas, 2005). By providing examples of biosocialities of people with a personal or family history of conditions ranging from Huntington's disease, to AIDS, to mental health disorders (Novas and Rose, 2000; Rose and Novas, 2005), they show that knowledge of one's own biology brings people together in new groups that construct a sense of belonging around a shared biological conception of themselves, or specific biomedical classifications.

Biosociality encourages activism in biological citizens who campaign for better knowledge of their condition, the development of new treatments, improved access to services and an end to stigma. In turn, activism leads to a reconfiguration of the relation between 'experts' and 'lay people', in which informed biological citizens demand control over practices related to their conditions and expect experts to support them to promote biotechnological progress relevant to their personal or family biological history (Novas, 2003). Because knowledge and beliefs about one's biology become part of complex choices that prudent individuals must make, every good citizen who accepts responsibility for their, their family and their group's wellbeing actively contributes to society's drive for health (Rose, 2001).

It is important to note that in their discussion about citizens' moral duty to actively participate in such drive for health, Rose and Novas have focused on biological citizens' responsibility to draw on *genetic* information to maximize their health and minimize their risk of illness. Indeed, while

they refer to *biological* citizenship and citizens, with some exceptions they tend to use the concept of responsibility in relation to genetics, hence favouring the expression '*genetic* responsibility' (Novas and Rose, 2000). In their view, because of the recent advances in genetics, 'one has long been responsible for the health and illness of the body, but now one must also know and manage the implications of one's own genome' (Rose and Novas, 2005, p. 441).

These reflections make two clarifications necessary in the concluding remarks of the present section. First, the emphasis placed on genetic information does not mean that Rose and Novas discuss genetic responsibility only in relation to people with a personal or family history of a genetic condition. The larger claims that they make about life in the biomedical age (s1.1) imply that every biological citizen falls under the category of the 'genetically at risk' person, which is one manifestation of the broader discourse around the rise of somatic individuality and biological citizenship (Novas and Rose, 2000).

Nor should the emphasis on genetics be interpreted as the alignment of Rose and Novas's account with the well-known argument about the 'geneticization' of societies, which Lippman proposed in 1991 as follows:

> *Geneticization refers to an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviours and physiological variations defined, at least in part, as genetic in origin […] Through this process, human biology is incorrectly equated with human genetics, implying that the latter acts alone to make us each the organism she or he is […] 'Geneticization' goes further, however, and poses genetics as the source of illumination itself [of social concerns], not merely one of the ways in which it might be refracted.*
>
> (Lippman, 1991, p. 19)

Rose and Novas have, in fact, criticized Lippman's perspective for interpreting recent advances in biology and genetics as leading towards an increase in genetic deterministic perspectives, and so towards individuals' passivity in relation to a hegemonic biology–destiny. Such an argument cannot fit the traditional notion of biological citizenship because, as shown above, in Rose and Novas's view biology has ceased to be a synonym of an immutable destiny that imposes limits on human

possibilities, to become amenable to control and choice (Rose, 2007; Rose and Novas, 2005). For instance, far from being definitive in most cases genetic information provides indication of a probability or risk (Rose and Novas, 2005). In this sense, ethopolitics is an ethics of *active* citizenship that does not lead to fatalism but encourages the patient-subject to take responsibility for maximizing their chances of health (Novas and Rose, 2000).

Moreover, although genetic attributions are central to processes of personal and group identity formation, and to citizens' development of a sense of self as moral and political agents, in contrast to Lippman's theory, Novas and Rose argue that '[i]deas about biological, biomedical and genetic identity will certainly infuse, interact, combine and contest with other identity claims' (2000, p. 491). Similarly, while biomedical knowledge has become a priority in the life of lay people as biological citizens, this is only one among many forms of expertise (e.g. experiential expertise), on which citizens ground risk calculations and choices about the future (Rose, 2007).

Rose and Novas's theory has had a huge influence on the literature on biological citizenship. However, as the traditional notion grew in popularity, so did the recognition of some of its shortcomings. These will be discussed in the next section, to show how they sparked the intellectual interest that motivated me to pursue the present research.

2.2.2. Challenging Rose and Novas's account

While reviewing the critical literature, I identified two major concerns about the traditional notion of biological citizenship. The first concern challenges Rose and Novas's assumption that in western democracies there exists a hegemonic understanding of subjectivity in biological terms, and that citizens of western democracies have come to endorse such a narrative; while the second criticism affirms that the particular kind of biological citizen constructed by Rose and Novas is not representative of any citizen of the biomedical age.

Critics tend to engage with Rose and Novas's arguments on a descriptive level, i.e. they question that the traditional notion of biological citizenship accurately reflects life in modern western democracies; however, their criticisms have important normative implications that interlace with broader ethical issues discussed within the disability studies and the scholarship on gen-ethics. Therefore, a critical review of the traditional notion could potentially address a literature of much wider scope. Because there is no space to do so in this chapter, the next sections focus primarily on those contributions from disability studies and the ethics of genomics that have more closely and explicitly engaged with the literature on biological citizenship.

2.2.2.1. A reductionist account of identity and citizenship

Some scholars have argued that Rose and Novas's description of how biological citizens relate to themselves and understand their selfhood provides a reductionist interpretation of identity, and consequently of citizenship. Reductionism here refers to their emphasis on biology and genetics which, as the critics have cautioned, only grasps one aspect of citizens' identity claims. To illustrate this point, scholars such as Plows and Boddington (2006) have referred to the debate in disability studies, concerning the way disability should be understood and society should relate to it.

A range of suggestions have been proposed in this debate. Until a couple of decades ago, the dominant perspective was that of the social model of disability. Particularly in its British version, which emerged in 1970s, the social model criticizes medical accounts that reduce disability to the biological mechanisms underpinning disabled people's condition and thus construct disability as a 'problem' of the individual body. The social model postulates a strong separation between 'biological impairment' and 'disability', and argues that while disabled people have a biological (physical or mental) impairment, it is society that disables people with impairments by taking little or no care of their needs (Oliver, 1996, quoted in Shakespeare and Watson, 2001, p. 10). Thus, disabled people's social disadvantage or their socially constructed disability is solely caused by societal barriers, i.e. by a structural and societal organization chosen by non-disabled people. By

rejecting a biological understanding of themselves and their own disability, social model activists oppose the use of biomedical knowledge as the means to fix a biological problem that is regarded as preventing them from adapting to the existing social structure. By highlighting the way in which social environments construct disabilities, they instead demand that it is society that should change, i.e. remove discriminatory barriers to accommodate disabled people's needs.

More recent developments within disability studies have led disability activist and scholar Tom Shakespeare to criticize the 'strong' social model which, he argues, overemphasizes the social creation of disability by devaluing the role that impairment plays in disabled people's lives. Shakespeare has pointed to several limitations that ultimately threaten the credibility of the 'strong' social model. These include the strict separation between individual/biological impairment and social/constructed disability; the lack of recognition that many impairments are intrinsic limitations that would still leave some (or most) disabled people at a disadvantage, even if societal barriers were removed; and the idea of a barrier-free world, which Shakespeare labels as 'utopia' (2014, p. 36), because provisions for achieving it for any one group of disabled people are impractical and likely to be incompatible with another group's needs.

Thus, Shakespeare and other scholars (see Danermark and Gellerstedt, 2004) have drawn the conclusion that a more accurate and robust theory of disability should shift away from the focus on 'the medical' versus 'the social', which are both reductionist models that foster counterproductive polarizations. They instead favour a complex and multifactorial account, which includes various causal explanations of disability and components of disabled identity (e.g. biological, social, psychological, cultural, socioeconomic, normative, political, etc.). Under this approach disability is 'the outcome of the interaction between individual and contextual factors' (Shakespeare, 2014, p. 77), and so 'people are disabled by society *and* by their bodies' (Shakespeare, 2014, p. 75). This model, it is argued, is best placed to account for and address the diverse experiences that disabled people have of their own disability (Hughes, 2009; Shakespeare, 2014).

Many more pages could be devoted to discuss this literature. However, for the purpose of a critical review of the concept of *biological* citizenship, it will suffice to note that the debate within disability studies has taught us that the processes through which people develop their identities, as individuals and citizens, are characterized by various layers of intricacy, which Rose and Novas's emphasis on the biological seems to have overlooked. As Plows and Boddington put it:

> *Someone diagnosed with a specific syndrome could identify themselves as a member of a patient group wanting cures or as a member of a disability rights group mobilizing over access to treatments and a range of social provision. Such an individual could see their syndrome as core to their identity, or could reject the centrality of a biologically informed construction of their own identity. They may hold, or feel strongly antagonistic to some of these positions, and/or ambivalent about some or all of them. The core issue here is to recognize the complexity and ambivalence surrounding any construction of identity and related citizenships, because identities are multiple and also fluid.*
>
> (Plows and Boddington, 2006, p. 123)

As presented above (s2.2.1), Rose and Novas's account allows the space for non-genetic and non-biological claims, and in this sense they have distanced themselves from the geneticization argument (Lippman, 1991). Thus, it could be argued that the first objection to the traditional notion does not do justice to their critical engagement with Lippman's theory. However, by definition reductionist positions do not deny that there may be non-biological or non-genetic explanations of a certain phenomenon, but these are seen as playing a minor role as background conditions, i.e. even if available they 'would not get to the heart of the matter' (Wachbroit, 2000, p.355). Therefore, Rose and Novas promote a reductionist perspective in the sense that they seem to argue that the biological interpretation is the most appropriate, or privileged, understanding of subjectivities and citizens in the current times. It is precisely this privileged understanding that the critics contest. By overemphasizing the biological dimension, Rose and Novas have not adequately engaged with the longstanding opposition that certain groups in society, and so some of those that they would call biological citizens, have raised against the growing biologization of physical and mental states.

2.2.2.2. An elitist conception of biological citizens

The second criticism of Rose and Novas's account regards the examples that they use to construct the profile of the biological citizen. Two main characteristics recur in these examples: first, biological citizens are empowered, educated, well-resourced individuals who are able to freely engage with the citizenship values promoted in the biomedical age; second, these individuals are motivated to construct a positive and favourable relation to biomedical knowledge and to the apparatus of biotechnology, and use this and the advances made possible by it to reach their goal of increasing their access to treatments, interventions and services.

Neither characteristic seems to convince the critics, who have discussed three kinds of biological citizens that do not align with Rose and Novas's description because they interact with biotechnology in alternative ways. In the next sub-sections, these will be presented separately for explanatory purposes, but their identities are intertwined and often coexist in the same individual. This review will highlight that the kind of biological citizen described under the traditional notion 'is not everyman' (de Vries et al., 2017, p. 18), and so that Rose and Novas's account is 'inherently elitist' (Plows and Boddington, 2006, p. 123) because, perhaps unintendedly, it excludes alternative ways of being a citizen of the biomedical age.

Citizens who are subjected to tacit forms of coercion

First, it has been argued that Rose and Novas have failed to account for 'the difference between citizens freely mobilizing in relation to biotechnology, and "citizens" actually being manipulated [by it]' (Plows and Boddington, 2006, p. 121). Even though the language of biomedicine and genetics seems to be offering choice, some authors have pointed out that in reality it imposes explicit or implicit 'pressures to be "prudent"' (Kong et al., 2017, p. 7), of which citizens may not be necessarily aware. In a culture where being healthy has become a moral value, these pressures often take the form of negative judgements directed towards those who make 'irresponsible' choices.

There seems to be abundant evidence of these tacit forms of coercion in various contexts of the application of genomic science. One example is Hallowell's sociological work (1999a, 1999b) on genetic counselling for women (potentially) at genetic risk for hereditary breast and ovarian cancer, which found that counselling is not neutral, but explicitly or implicitly prescriptive. Counsellors tend to describe the risk of cancer as manageable and, in order to encourage women to undertake risk-management practices, they present the various options available to their clients selectively, by either avoiding to discuss the option of not pursuing further medical interventions, or by presenting it as irresponsible.

More recent evidence indicates that similar forms of implicit or explicit pressure have arisen in the context of reproductive genetics (Biesecker, 2019; Johnston et al., 2017), in relation to genetic tests such as prenatal screening (PNS), prenatal diagnosis (PND), preimplantation genetic diagnosis (PGD), and non-invasive prenatal testing (NIPT) (Box 1).

Box 1: Genetic technologies for reproductive choices

- **PNS**: It identifies women with an increased chance of having babies with certain chromosomal abnormalities. Women who receive a positive result indicating that the chance is higher than a certain cut-off are offered more testing (Genetic Alliance; The New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services, 2009).
- **PND**: It detects changes in a foetus's genes or chromosomes before birth. It is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. A test result can lessen a couple's uncertainty, or help them make decisions about whether to continue or terminate the pregnancy. However, PND cannot detect all possible disorders or defects (Genetic Alliance; The New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services, 2009).
- **PGD**: It is performed on embryos produced via assisted reproductive techniques from couples who are at risk of transferring a genetic condition to their offspring. The aim is to test *in vitro* whether the embryos carry any genetic mutations associated with the condition so to implant only those without such mutations in the uterus. PGD helps people to have offspring free of the condition without having to go through PND during pregnancy and subsequent termination of pregnancy (National Library of Medicine (US). Genetics Home Reference, 2020e).
- **NIPT**: It determines the risk of a foetus being born with genetic abnormalities, most often those caused by an extra or missing copy of a chromosome, by analysing the foetal DNA fragments circulating in a pregnant woman's blood. Being 'non-invasive', it poses fewer

risks to the foetus than invasive tests (National Library of Medicine (US). Genetics Home Reference, 2020f).

Johnston et al. (2017) have pointed to the evidence from the US showing that women who undergo prenatal genetic screening and diagnostic tests often have little understanding of the kind of information that tests will produce and of its implications. Lack of time and professionals trained to provide comprehensive counselling sessions to expectant parents has indeed resulted in the routinization of many tests done prenatally, and so consultations often become the responsibility of primary prenatal care providers, who 'can be quite directive […] about prenatal testing, perhaps owing to their perceptions of the value of such information and the emphasis of their training' (Johnston et al., 2017, p. 506).

This evidence confirms Shakespeare's argument (2014) that two factors play a role in undermining couples' opportunity for making choices about the use of reproductive genetic technologies in a way that is as free and fully informed as clinical rhetoric presents it, and as Rose and Novas have assumed. There are conditions that apply to couples' discrete experiences of pregnancy services, in which information provided about prenatal genetic technologies seems to be biased towards the experience of being tested and the technical details of the test (e.g. reliability). A balanced discussion about the conditions that could be detected, and what it would mean to raise a child with those conditions, is often neglected, which may be interpreted by parents as suggesting that any detectable condition should be prevented.

This is intertwined with the second factor, which pertains to the broader cultural context, particularly societal knowledge of and attitudes towards parenting and disability. Across countries, there seem to be widespread professional and lay assumptions about what constitute (un)desirable pregnancy outcomes (Landsman, 2009; Remennick, 2006). This implies that were parents to make 'irresponsible choices' (e.g. opting for an unmediated, at-risk pregnancy), many would be exposed

or would at least perceive to be exposed to negative societal judgements. Indeed, prospective parents (especially mothers) often consider becoming informed about their future children's potential genetic risks as part of good and responsible parenting (Roberts, 2010). Thus, it is not surprising if a recent systematic review of qualitative and socio-empirical work on genetic responsibility identified that in many studies participants valued autonomous decision-making about tests, but were sceptical of the possibility of achieving it in practice (Leefmann et al., 2017); 'responsible' choices can indeed be determined by the responsibilities people feel towards broader cultural, social, and moral norms.

Drawing on this literature, one cannot but recognize the unjustified optimism that characterizes quotes such as the one below, in which Novas and Rose seem to understand biomedical practices primarily as empowering already educated and well-resourced individuals who are capable of occupying a dominant position within the biomedical apparatus, and whose choices are therefore autonomous, free and informed:

> *[S]uch persons […] are increasingly demanding control over the practices linked to their own health, seeking multiple forms of expert and non-expert advice in devising their life strategies, and asking of medics that they act as the servants and not the masters of this process.*
>
> (Novas and Rose, 2000, p. 490)

It is because of this overly positive interpretation that Novas and Rose's framework cannot account for the tacit forms of pressure that exist in the age of biomedicine. Critics conclude that their construction of a harmonic relation between citizens and biomedicine could end up serving the interests of the market of biotechnologies (Plows and Boddington, 2006).

Citizens who reject biotechnological advances linked to their condition

The second group of people who do not align with Rose and Novas's description of the biological citizen includes those who reject the technologies meant to prevent, treat or eradicate their or their loved ones' conditions. Ultimately, these individuals overtly oppose at least some of the citizenship

values that are promoted under the traditional notion and drive biotechnological advances. A paramount example of these attitudes is offered by disability activists, whose rejection of biotechnology follows from their uneasiness with a purely biological definition of disabilities (s2.2.2.1):

> *While health activists often make demands for increased access to medical resources, disability activism is frequently characterized by an opposite strategy: escape from medical definitions to an insistence on political and civic entitlements has produced a distinct and potent agenda.*
>
> (Heath et al., 2008, p. 158)

Since the progress made at the time of the Human Genome Project, disability activists and scholars have contended that increased understanding of the biological underpinnings of disabilities will lead to the development of applications to ultimately eliminate their disabilities. Their concerns have focused on prenatal tests (PGD and PND) that identify biomarkers for various disabilities in embryos or foetuses with a view to terminating affected pregnancies – a debate that has been recently reignited by new developments, such as NIPT (see Valentin et al., 2019) and gene editing (see Parens and Johnston, 2019). Criticisms have also been directed towards population newborn screening to identify infants who are affected by, or at risk of, certain conditions, as well as towards pharmacological and other biological interventions that are meant to fix 'biological defects' later in life, such as hearing aids or cochlear implants for people with hearing loss (Sparrow, 2005).

Many scholars, including bioethicists, have written about the 'suspicion' (Shakespeare, 2014, p. 18) with which at least part of the disability movement regards biotechnological interventions attempting to mitigate or cure people's impairments (see Buchanan et al., 2000; Roberts, 2010; Savulescu, 2002; Scully, 2008; Scully, 2019). Some empirical studies have been conducted with relevant stakeholders, including people with or caring for individuals with achondroplasia, deafness and Down syndrome (see Burton et al., 2006; Gollust et al., 2003; van Schendel et al., 2016) – in addition to autism, as I will discuss in Chapter 3. Clearly, these multiple and heterogeneous disabilities raise concerns with different nuances (Scully, 2008). However, the disability

movement's suspicion can be summarized around three main arguments, which signal a common underlying concern that the most recent improvements in genetics and biotechnologies might reinstate the eugenic aspirations of the last century (Buchanan et al., 2000).

First, it is argued that, as the number of disabled people drops because of the availability of preventative or therapeutic interventions, societal support and social provisions for people who already have those disabilities will decrease. Second, people with hearing loss who identify with the Deaf culture argue that some disabilities have allowed the rise of new cultures, i.e. minority groups with shared language, identity, history, values and so on. Not only do they resist being identified as disabled groups, and so reject technologies such as cochlear implants to 'fix' them, they also highlight the benefits that members reap from being part of these cultures, which may counterbalance or even outweigh the disadvantages deriving from having the impairment. Thus, they challenge appeals to the principle of beneficence to support the moral responsibility to draw on biological and genetic information for the prevention or treatment of impairment. Finally, the 'expressivist argument' affirms that efforts to develop interventions that use biomedical knowledge for prevention and/or treatment are grounded on ideas that express negative judgements towards disabled people and discriminate against them by devaluing their lives.

There is no space here to exhaustively explore these arguments and the objections that they have stimulated, nor is this the aim of this section; in fact, the next chapters will offer the opportunity to expand on some of these considerations by using autism as a case study. However, this brief illustration suffices to demonstrate that, in contrast to what is suggested by Rose and Novas's examples of biosocialities, not all citizens of the biomedical age welcome biomedical interventions and construct a sympathetic relation with the apparatus of biotechnology. Disability activists' claims have shown that some citizens build an antagonistic relation to medical advances and practices related to their health conditions. This is because they reject the idea of a citizen that is valued within the traditional notion of biological citizenship, i.e. one who acts responsibly if they

use genetic knowledge to minimize the risk of traits that society considers undesirable. This consideration introduces the third and final group who escapes the categorization of biological citizens under the traditional notion.

Citizens whose values differ from those of the moral framework of biotechnology

The last group is composed of individuals who neither enthusiastically support the endeavour of biomedicine, nor make their opposition to it part of their group identity. On the contrary, their choices show that they prioritize a different set of values than those of the moral framework underpinning biological citizenship projects. This alternative kind of biological citizen usually emerges when people are confronted with choices about genetic testing in reproductive contexts (PGD and PND), and perinatal care (newborn screening or NBS). These choices indeed encourage parents to think of their understanding of good parenthood and their visions of the family they would like to raise. Two recent studies explicitly engaging with the literature on biological citizenship illustrate this point.

In the first study from the US, Raspberry and Skinner (2011) investigated the reproductive choices and enactment of responsibility of women who carried the gene for Fragile X (FX) syndrome, and so have a 50% chance per pregnancy of passing on their gene, and who already had the experience of parenting one child with the condition. While most participants decided not to have more biological children, a significant minority either purposely initiated an unmediated pregnancy, or continued an at-risk unplanned pregnancy. In the authors' interpretation, the former group's understanding of responsibility was close to Rose and Novas's account of 'managing genetic risk' (Raspberry and Skinner, 2011, p. 424). The latter group instead understood responsibility as the duty to 'love, care for, and for some, procreate all children, regardless of their genetic status' (Raspberry and Skinner, 2011, p. 424).

In the second study, Raz et al. (2019) found that similar concerns and underpinning values were expressed in the context of NBS, which consists of pricking newborns' heels to collect a few blood

drops and test them for a range of disorders. The authors interviewed parents whose children had tested positive to a range of conditions screened for through NBS in Israel. Because in some cases NBS could affect future reproductive choices, particularly when parents come to learn they are carriers of the genes for which their children have screened positive, participants were asked about their attitudes towards the use of PND, should they (or their partner) become pregnant again.

Only a minority of interviewees affirmed that they would undertake PND to eventually terminate their pregnancy because of reasons related to family hardship in raising another affected child. The majority of participants (53%), all with children with relatively milder symptoms, reported that in subsequent pregnancies they would not access prenatal genetic services, and 21% would only do so to be prepared for the birth of another affected child. Although hypothetical and prospective interest does not always appropriately predict uptake of genetic testing (Sanderson et al., 2010), these findings suggest that experiences of raising a child with a genetic condition influence the way in which parents understand genetic responsibility. In some cases such experiences translate into acceptance of existing children's diversity and openness towards raising more children with the same health difficulties (Raz et al., 2019).

There are plenty of other studies on parents' views of and attitudes towards the use of genetic technologies in prenatal and perinatal care, and any review of this literature in the context of the present chapter will never do justice to the complexity of the ethical issues at stake. Not only could the results of PGD, PND and NBS raise slightly different normative concerns because they are performed at different life stages. Some have also argued that different conditions may inform different attitudes (Hayeems et al., 2008), and there seems to be cultural variation in the way these technologies are perceived (Raz and Schicktanz, 2009). Furthermore, two recent systematic reviews have shown that investigations of the use of genomic information about carrier status ascertained through NBS for future pregnancies give mixed results (Hayeems et al., 2011), and that

in studies about reproductive genetics the notion of responsibility is deployed in relation to contrary outcomes of decisions, e.g. to abort as well as not to abort (Leefmann et al., 2017).

This complexity demonstrates that not all parents understand responsibility in the simplistic sense attributed to it by Rose and Novas, i.e. as a duty to know about and act on their (future) children's genetic status. On the contrary, some parents operate within a framework in which the availability of genetic technologies allows another sense of responsibility to emerge, in which being a good parent consists of valuing parenthood, celebrating the family, and accepting and honouring a child's specialness and difference (Raspberry and Skinner, 2011). Thus, this literature highlights 'the diversity of normative scripts of "genetic responsibility" in the context of parenthood' (Raz et al., 2019, p. 48) or 'the complex and situationally-dependent aspects of genetic responsibility' (Raspberry and Skinner, 2011, p. 422). In this sense, the realities in which people make decisions about PND, PGD and NBS are more nuanced and complex than Rose and Novas have admitted.

The reasons why some parents do not align with Rose and Novas's theory are multiple, but here I want to draw attention to two considerations. First, both studies described above suggest that the experience of having a child with a certain condition often challenges parents' assumptions about what they had considered as a normal or healthy child, and it makes new kinds of 'imperfect normality' emerge (Buchbinder and Timmermans, 2011, p. 76). Second, in a paper discussing parental views about the use of whole genome sequencing in newborns, Johnston and Juengst have suggested that parents may decline certain tests because 'when they have a chance to think through the option in conversation with a clinician, they come to understand the low odds of discovering anything that they can do something about' (2018, S15).

This does not apply only to information that is not medically actionable. Most conditions have complex (i.e. non-Mendelian) inheritance (Torkamani et al., 2018), and their underlying genes are characterized by incomplete penetrance and variable expressivity (Box 2 below); consequently, among those who carry the genes associated with these conditions, it is difficult to predict who will

develop them, as well as their degree of severity. Furthermore, the interpretation of the genetic information that tests retrieve has *per se* become more complex as a consequence of the shift from the study of single genetic mutations to the study of the entire human genome (s1.1). Whole genome sequencing can identify variants of unknown significance (VUS), i.e. genetic mutations whose function has not yet been established. Thus, some test results could provide information that has little clinical validity and clinical utility (Box 2), i.e. information about which parents cannot do anything, or with which they would not know what to do.

Box 2: Genetic penetrance and expressivity, and clinical validity and utility of genetic tests

- **Penetrance**: The probability that an individual who has inherited a genetic predisposition to a certain condition will manifest the signs and symptoms of that condition. If such probability is lower than 1.0, the genetic predisposition has incomplete penetrance (National Library of Medicine (US). Genetics Home Reference, 2020b).
- **Expressivity**: Most genetic conditions cause signs and symptoms that vary among affected people. Variable expressivity refers to the range of such signs and symptoms that people with the same genetic condition manifest (National Library of Medicine (US). Genetics Home Reference, 2020b).
- **Clinical validity**: 'It refers to how well the genetic variant being analysed is related to the presence, absence, or risk of a specific disease' (National Library of Medicine (US). Genetics Home Reference, 2020a).
- **Clinical utility**: 'It refers to whether the test can provide information about diagnosis, treatment, management, or prevention of a disease that will be helpful to a consumer' (National Library of Medicine (US). Genetics Home Reference, 2020a).

This may explain why some empirical studies report that the availability of genetic testing does not necessarily correlate with people's desire to use it in light of a heightened sense of genetic responsibility towards others (Leefmann et al., 2017); or that some parents may *responsibly* decide to decline certain tests and focus on other issues that may arise during pregnancy or in their children's first days of life (Johnston, 2018; Johnston et al., 2017; Johnston and Juengst, 2018).

The considerations about the increasing layers of complexity that characterize choices about the use of genetic information in the genomic era ties well into reflections of what biological citizenship means in mental health, as I will discuss in section 2.3.

2.2.3. Building on the critical literature

Although in Chapter 1 I argued that Rose and Novas's account is still relevant to the current times, the above considerations have highlighted some important limitations of their traditional notion of biological citizenship. Critics have pointed out that citizens do not necessarily subscribe to a biological understanding of themselves and their identity, which Novas and Rose consider to be dominant in the age of biomedicine. Moreover, biotechnology does not necessarily create *subjects*, i.e. citizen-consumers who develop an active, rather than passive, relation to biomedical advances; nor do all citizens positively welcome biomedical practices linked to their own health or embrace the values underpinning efforts to make these advances possible. On the contrary, in addition to the kind of citizens described by Rose and Novas there are people who (perhaps unknowingly) are subjugated to the interests of the apparatus of health; who are against biotechnologies that are meant to predict, prevent, treat and eventually eradicate their condition because they reject the value judgements embedded in the moral framework of biotechnology; and those who, despite not being explicitly opposed, make choices that reveal their endorsement of a different set of values.

I argue that these criticisms emerge from three assumptions underpinning the traditional notion of biological citizenship that seem unconvincing and so warrant further investigation:

1. *Alleged consensus over the distinction between health and illness:* Rose and Novas have argued that biology has become manipulable, which implies that risk to pathological states can be acted upon to produce better (i.e. healthy) life outcomes. However, the obligation to 'maximize health' and 'minimize the risk of ill health' seems to be grounded on the assumption that in the age of biomedicine an agreed-upon distinction exists between health and illness, or that agreement can be reached about what constitutes pathological and normal states. The aforementioned claims of disability rights activists and scholars have demonstrated that this is not necessarily the case. Although most people would consider disabilities as problems to be 'fixed' by medical expertise, some disabled individuals reject

the application of biological categories to their disabilities, oppose biomedical technologies that aim to cure them and see their conditions as differences that enrich the diversity of human existence. Thus, because Rose and Novas have chosen examples of biological citizens who construct a positive relation to biotechnologies, they have failed to account for cases in which underlying disagreements around the meaning of 'pathology' and 'normality' exist, and to recognize that tacit forms of pressure around life optimization and risk management may have unintended consequences in terms of shaping societal ideas of normality and pathology.

2. *Linear relationship between genetic risk and action:* The strong emphasis on genetic responsibility and citizens' obligations to use biological information to make choices and calculations in the present in light of their susceptibility to diseases in the future seems to assume a linear relationship between knowledge acquisition and action, or between information about genetic risk and life choices. This does not mean that Rose and Novas's theory is based on genetic deterministic assumptions, against which they have in fact cautioned. However, the ethics of active citizenship and the risk-management model that it adopts are arguably based on a circumscribed notion of genetic risk and of its potential for providing actionable information. The above discussion of the ethics literature on the use of genetic tests in prenatal and perinatal care has indeed shown that choices concerning these technologies are not only based on awareness of genetic risk. On the contrary, they involve 'a complex negotiation of personal desires, family values and diversity, religious faith, and financial constraints' (Raspberry and Skinner, 2011, p. 430). The same literature has also highlighted that our current technological ability to acquire complex information about people's biological life means that in many circumstances it has become increasingly complex to interpret genetic risk and to know what to do about it. Hence, if the availability

of information about people's biology and biological predispositions does not lead to fatalism, it may at least cause a paralysis of action.

3. *Conformity among duties, choices, and values:* Rose and Novas have developed a conception of citizenship in which individuals' choices, and the values underpinning them, are aligned with what is expected of good members of societies, i.e. to use genomic information to maximize health outcomes, and in which citizens are always in a position to act on such duty. However, the studies discussed in s2.2.2.2 have highlighted that, although citizens may not be aware of it, in some settings responsibilities have become 'crypto-duties' (Plows and Boddington, 2006, p. 127), i.e. they masquerade as choices. Moreover, from points 1 and 2 above it should be concluded that not all citizens align with the moral framework lying behind such duties and responsibilities. This means that to put all individuals in the same category of biological citizens is misleading, because such a broad definition masks a wide range of (often opposing) experiences, values, interests and ends. In this sense, the traditional notion fails to account for those members of society who do not match the profile of the neoliberal citizen-subject.

These three considerations are not meant to say that Rose and Novas's view is necessarily wrong, but that there are reasons to contest its applicability to account for life in the biomedical age beyond the examples of biosocialities that they have provided. In the remainder of this chapter, I will show how the shortcomings highlighted above become manifest in relation to the twofold focus of this thesis, i.e. mental health settings and young people. In section 2.3, I will illustrate the various layers of complexity that characterize the field of psychiatric genomics to question whether assumptions 1 and 2 can still be considered defensible given the most recent advances in genomics. Section 2.4 will argue that the peculiar condition that young people as 'genomic citizens in the making' (Sabatello and Appelbaum, 2016) hold in society means that they may add to those social groups for whom a false harmony between choices, duties and values is constructed (assumption 3).

These arguments will be pulled together in section 2.5, where the chapter concludes that, in light of the unprecedented effort recently put on psychiatric genomics, as well as of the increasingly recognized importance for the younger generations to contribute to matters of societal relevance, an empirical study should be conducted to put the traditional notion of biological citizenship to the test both in psychiatric settings and from the perspective of young people. Thus, in this thesis I build on the existing critical literature by investigating the meaning that *young people* attribute to the notions of biological citizenship and genetic responsibility in the context of *mental health*.

Although the literature on genetic responsibility has shifted from discussions focused on monogenic to multifactorial conditions after 2011 (Leefmann et al., 2017); and despite Rose's reflections on the application of his account to mental health, this will be the first study to provide a thorough investigation of biological citizenship entirely focused on psychiatry. Notably, it will also be the first investigation of these issues to involve young people.

2.3. Citizenship and psychiatric genomics

As one of the leading experts on the sociology of psychiatry Rose has argued that one specific manifestation of the somatic individual is the 'neurochemical self' (Rose, 2003). This emerged in the second half of 20th century as a result of thoughts, wishes, emotions, desires and conducts being understood and described with reference to neurochemicals and, subsequently, to the genetic and genomic variants underpinning neurotransmitters (Rose, 2007). According to Rose, in mental health biological citizenship often takes a specific connotation because of critical- and anti-psychiatry movements, which reject the genetic aetiology of psychiatric conditions and any attempts to 'fix' these through chemical treatments. Rose argues that, in contrast to parents of children with mental health problems, who may exercise traditional forms of active citizenship by seeking biological information to address their children's issues, some patients support anti-psychiatry movements by condemning reductionist applications of the biomedical model to

psychiatry, which they criticize as ultimately aiming to depersonalize and govern those with mental health issues (Rose, 2007).

However, except for those passages focused on critical psychiatry movements, Rose seems to have aligned his conception of neurochemical citizenship to his broader discussion of the ethics of active citizenship. By providing examples of psychiatric patients who commit to become experts on their conditions, and demand control over the medical practices linked to their care, he has argued that in psychiatry too patients have become skilled, prudent and active, hence concluding that:

> *The forms being taken by contemporary neurochemical selfhood […] are intrinsic to the continuous task of monitoring, managing, and modulating our capacities that is the life's work of the contemporary biological citizen.*
>
> (Rose, 2007, p. 223)

I argue that this, coupled with the fact that Rose and Novas's examples of biological citizens are as divergent as people with HIV/AIDS and people with bipolar affective disorder (Rose and Novas, 2005), suggests that they have not extensively engaged with a range of peculiarities that characterize the context of mental health. In particular, two main features are central to the discussion about the ethics of psychiatric genomics, and so warrant thorough investigation. These are various uncertainties that characterize psychiatric conditions, and the stigma attached to them (Figure 3).

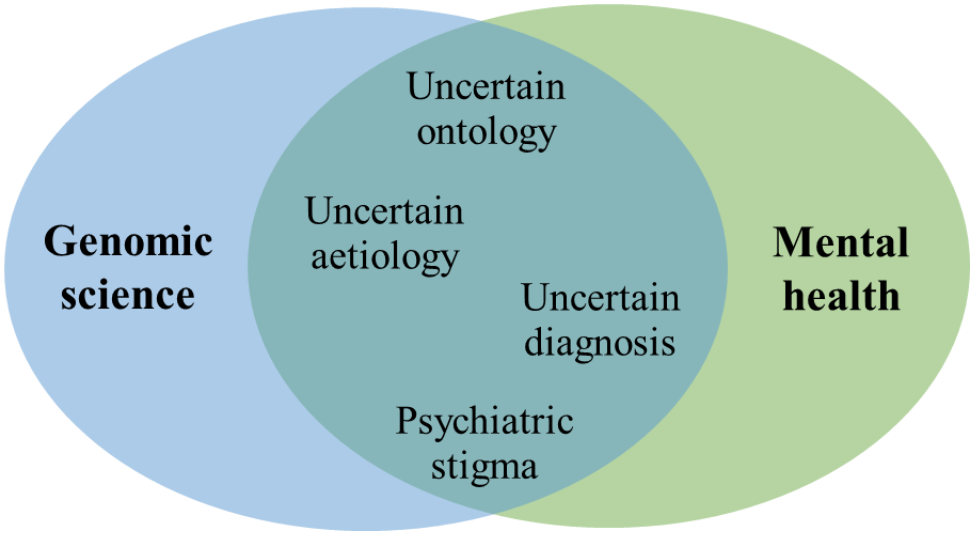


Figure 3: The various layers of complexity that emerge from the application of genomic science to mental health

In the next pages, I will show that the uncertainties characterizing psychiatric conditions raise questions about the purpose of neuropsychiatric genomics, i.e. whether we *can* and *should* predict, prevent and treat behavioural traits that are cast as 'deviant'.[9] The fact that psychiatric conditions are highly stigmatized poses questions around what kind of attitudes and value judgements about psychiatric disorders and people suffering from them ground societal demands for using genomic information to predict, prevent and treat psychiatric conditions.

Of note, I do not aim to argue that these features are unique to conditions that affect behaviour (i.e. psychiatric disorders) as opposed to somatic or physical diseases and disabilities. Similar complexities emerge from the application of genomics to disorders such as hearing loss, which is a common and complex condition characterized by disagreement over the value judgements that should be attached to it. However, drawing a thorough comparison between different conditions is

[9] Because of the complex biological underpinnings of psychiatric conditions, Rose's most recent work (Rose, 2018) is imbued with scepticism over the optimistic discourse that many psychiatric geneticists still propose about the future possibilities of the field. However, this is not discussed in relation to the theory of biological citizenship, which further demonstrates that such theory has not engaged in depth with the complexities that arise from the application of genomics to mental health.

beyond the scope of this research, which focuses on mental health. By discussing a range of psychiatric uncertainties and reflecting on psychiatric stigma, I intend to derive some implications on the traditional account of biological citizenship, and so question some of Rose and Novas's assumptions concerning life in the biomedical age.

2.3.1. Ontology, aetiology and diagnosis: Various layers of psychiatric uncertainty

A considerable amount of literature has been published on the ethics of uncertainty in relation to technological advances (see Sollie, 2007; Tannert et al., 2007). In this section, I will focus on two approaches that help to shed light onto the discussion of uncertainty at the intersection between genomics and psychiatry.

Han et al. (2011) distinguish between *sources* and *issues* of uncertainty. They argue that uncertainty may derive from the indeterminacy of a phenomenon or of the outcomes of a certain intervention ('probability'); the limitation in knowledge of a certain phenomenon ('ambiguity'); and the perceived incomprehensibility of a piece of information characterized by some features that make it hard to understand ('complexity'). These sources of uncertainty may contribute to different issues of uncertainty, two of which are relevant to the present discussion. First, 'scientific' uncertainty encompasses 'uncertainties about diagnosis, prognosis, causal explanations and treatment recommendations' (Han et al., 2011, p. 833). Second, 'personal' uncertainty concerns the existential and psychological impact that a certain condition might have on one's wellbeing, relationships and life goals.

The second approach was developed by Babrow et al. (1998), who discuss five *forms* of uncertainty, which overlap with Han et al.'s taxonomy, but have a more specific focus on individuals' personal experiences. Building on these theories, bioethicist Ainsley Newson and colleagues have defined uncertainty in relation to genomic testing as follows:

> *Genomic uncertainty is a status quo that arises when information that is obtained from genomic testing is imperfect or unknown, leading to uncertainty in clinical diagnosis or*

> *management. Genomic uncertainty can arise from the probabilistic, ambiguous or complexity uncertainty inherent to the information arising from testing, or from the provider's or recipient's views on and uses of it.*
>
> (Newson et al., 2016, para. 10)

I believe that Newson et al.'s definition is helpful to understand three intertwined forms of uncertainty that emerge in psychiatric genomics in relation to the following dimensions:

1. The ontology of mental disorders – What are they and why are certain behaviours conceptualized as 'disordered'?
2. The aetiology of psychiatric conditions – What causes the development of these conditions?
3. The diagnosis of mental disorders – Is there evidence for their validity and reliability?

Conflicts around these questions started long before identifying psychiatric biomarkers was even possible; in fact, as mentioned in s1.1, research on psychiatric genomics is motivated by an attempt to overcome problems that emerge from diagnoses that are based on behavioural symptoms (Rose, 2018). However, such an attempt has paradoxically fed into psychiatric uncertainty.

Uncertainties about the ontology and aetiology of mental health conditions are *scientific* issues, in Han et al.'s sense (2011), deriving from the 'probabilistic' and 'ambiguous' nature of genomic information. This in turn explains the 'complexity' that practitioners and patients may perceive in understanding the role of genomics in the ontology and aetiology of mental health issues, and it contributes to the uncertainty around developing valid and reliable psychiatric diagnoses. Uncertainty about mental health diagnoses also has a *personal* dimension, because it relates to the lack of consensus on the value judgements that should be applied to the existential, psychological and life outcomes of behavioural traits that some consider as deviant. It is such lack of consensus that explains the uncertainty that characterizes a 'provider's or recipient's views on and uses of' genomic information in mental health settings (Newson et al., 2016, para. 10).

2.3.1.1. Ontological and aetiological uncertainties

To understand the uncertainties concerning the ontological status and aetiology of mental health conditions a broader look at the longstanding debate about the meanings of 'health', 'disease' and 'disorder' is necessary. Although the literature on this topic is too vast to be thoroughly discussed here, it will suffice to say that some scholars have argued that, in western medicine discourses, the prevalent notion of 'disease' and 'disorder' is an ontological one (Jutel, 2011; Rose, 2018), which means that diseases are understood as 'ontologically real and definitionally specific' entities (Rosenberg, 2002, p. 239). In psychiatry, since the publication of the DSM-III in 1980, this model has inspired a categorical understanding of diagnoses. For decades the idea of mental health diagnoses as categories indicating an underlying discrete, homogeneous and separate entity with a specific aetiology, symptomatology (i.e. a set of 'abnormal' behaviours) and prognosis, has prevailed (Rose, 2018).

However, such widely used conceptualization of disease does not fit psychiatry well,[10] and a further exploration of the aetiology of mental health conditions will help to explain why. Psychiatric disorders do not seem to have clear boundaries on both a genotypic level and, for reasons that will become clearer below, a phenotypic level. Indeed, they are among the common and complex disorders that are caused by genetic variants with incomplete penetrance and variable expressivity (Box 2, s2.2.2.2). In particular, two kinds of variants seem to act in concert, and together with the environment, in the development of mental health conditions (Gelernter, 2015; Gershon and Alliey-Rodriguez, 2013; Schulze and McMahon, 2018). These are single-nucleotide polymorphisms or SNPs, and copy number variations or CNVs (Box 3).

[10] I am not arguing that psychiatry is unique in this respect (see for instance Kerr, 2004), but that this is a feature of psychiatric conditions that needs to be taken into account to understand a range of complexities that emerge when one tries to apply the 'traditional notion' to mental health settings.

Box 3: Genetic variants involved in the development of psychiatric conditions
SNPs: These are changes at a single-nucleotide level that are tested for in genome-wide association studies (Gelernter, 2015). Being present in > 1% of the population, they are considered common variants; however, taken individually SNPs only increase the risk of a certain condition of a small percentage (National Library of Medicine (US). Genetics Home Reference, 2020d). This explains why psychiatric genomics studies identify several genetic variants assumed to increase the risk of the psychiatric condition analysed. For instance, the Psychiatric Genomics Consortium has recently identified 128 independent schizophrenia-associated SNPs in 108 genetic loci (Schizophrenia Working Group of the Psychiatric Genomics Consortium, 2014). SNPs seem to also impact individuals' response to certain drugs and their susceptibility to environmental factors. **CNVs:** These are rare large chromosomal microdeletions and microduplications; i.e. they are mutations involving several genes and causing the individual to have one extra (or one less) copy of those genes (Gershon and Alliey-Rodriguez, 2013). They are detected by either targeted sequencing or whole genome and exome sequencing, and they can be inherited or arise *de novo* in an individual, if they are not present in the DNA of the individual's parents. CNVs are highly but not completely penetrant; i.e. compared to mutations like SNPs they significantly increase the risk for the condition they are associated with, and yet not all people who carry them will develop the condition (Gershon and Alliey-Rodriguez, 2013).

The lack of clear biological and genetic boundaries between different mental health conditions is due to the fact that some of these mutations increase the risk for various psychiatric diagnoses. For instance, 16p11.2 duplication, a widely studied CNV, contributes to the development of autism, schizophrenia and bipolar disorder (Shane et al., 2009). Furthermore, some variants are shared between affected and unaffected individuals, which makes it difficult to distinguish between pathogenic and benign ones, or to explain why they are disorder-provoking only in some individuals (Sebat et al., 2004). In light of this evidence, the field of psychiatry has come to recognize the dimensionality of psychiatric conditions, which are no longer seen as specific and discrete entities, but as manifesting across a continuum of function and impairment (Rose, 2018). In turn, this has stimulated efforts to re-conceptualize diagnosis in psychiatry, although so far this paradigm shift has been more visible in research than in clinical practice.[11]

[11] See for instance the Research Domain Criteria (RDoC) initiative launched in 2013 by the National Institute of Mental Health (Insel, 2013). For a critical discussion of this model, see Rose, 2018, Chapter 4.

Thus, the uncertainty surrounding the ontology and aetiology of psychiatric conditions seems to derive from the 'ambiguous' and 'probabilistic' nature of psychiatric genomic information (Han et al., 2011). These considerations suggest that Rose and Novas's circumscribed notion of genetic risk, which is presumed to always provide actionable information (assumption 2, s2.2.3), is unlikely to be applicable to psychiatry, and thus that the ethics of active citizenship is difficult to abide by in these settings. This is because of our currently limited knowledge of the genomics of mental health conditions; and due to the indeterminate impact that variants with incomplete penetrance and variable expressivity have on the phenotypic level, particularly when SNPs are taken individually.[12]

2.3.1.2. Diagnostic uncertainty

Some scholars have also pointed to the uncertainty over psychiatric diagnoses both over time and across cultures, societies and even clinicians, by questioning their validity and reliability. Validity refers to the accuracy with which certain diagnostic criteria identify a certain psychiatric condition and differentiate it from other disorders; reliability indicates the likelihood that different psychiatrists will consistently diagnose a certain patient with the same condition, or that the same psychiatrist will give a patient the same diagnosis at different times (Feighner and Herbstein, 1987). Several hypotheses could explain the uncertainty surrounding psychiatric diagnoses, some of which interlace with the above considerations about how to understand the ontology and underpinning biology of psychiatric conditions.

Like somatic disorders, conditions affecting behaviour are defined in terms of their deviance from a criterion of normality. Unless one adopts a (contested) biostatical theory that understands normality as a biological, statistical and value-free concept (Boorse, 1977), it could be argued that

[12] This is why researchers are investigating the clinical utility of polygenic risk scores (PRS), which sum individual SNP effect into single scores, for risk stratification and prevention, differential diagnosis and treatment stratification (Binder, 2019, Zheutlin et al., 2019), although ethical concerns have been raised (Palk et al., 2019).

in mental health settings the criterion of normality is a value judgement determined, at least in part, by what is considered as desirable behaviour within a certain society. This suggests that criteria of normality, and therefore of deviance from it, are context- and culture-dependent (Rosenberg, 2006).

Because societal and cultural values and beliefs change over time, so do diagnoses that are rooted in them. In western medicine, homosexuality, masturbation and some forms of female sexuality were once thought of as deviant (Rosenberg, 2002). Similarly, societal sensitivity towards certain behavioural traits that are still part of the DSM (e.g. autism, bipolar disorder, etc.) may be changing thanks to self-advocates who are publicly taking pride in the strengths that their mental traits have provided them with (Higashida et al., 2013; Newlyn, 2018). Correspondingly, much has been written about what some see as an increasing medicalization of mental states that were once considered as part of common human experience (Rosenberg, 2006), such as the re-conceptualization of grief as part of major depressive disorder in the DSM-5 (Pickersgill, 2014).

In addition, diagnostic labels are broad categories including heterogeneous symptoms, which are often shared among diagnoses. On the one hand, this means that individuals with a range of different clinical signs of different severity are often diagnosed with the same psychiatric condition. The broadening of diagnostic criteria in psychiatry has led some to question whether people with milder symptoms of, for example, ASD should really be treated within the medical system (Graf et al., 2017; Timimi et al., 2011). On the other hand, since the 1970s research studies have demonstrated that the heterogeneity of behaviours targeted by psychiatric diagnostic systems poses challenges to the probability that psychiatrists will reliably diagnose mental conditions (Rose, 2018).

Problems of reliability are worsened by the pragmatic and social function that psychiatric diagnoses have acquired, e.g. they grant support for individuals and families or insurance reimbursement (Rose, 2018; Russell et al., 2010). Rather than faithfully following the criteria outlined in accepted diagnostic systems, psychiatrists often assign the diagnosis that they believe is most helpful for

their patient given their social context, which may differ from the label that others showing the same symptoms would receive elsewhere (Grinker, 2008). Thus, it is unsurprising that people often receive multiple diagnoses or see their dominant diagnosis change overtime (Timimi, 2014).

Critics claim that in psychiatry diagnoses are uncertain (i.e. they have low validity and reliability) because they have not been linked to any 'objective' and 'stable' underlying biological mechanisms or markers, which is in turn due to the probabilistic and ambiguous nature of psychiatric genomic information outlined above.[13] This is why, despite unprecedented efforts in psychiatric genomics, some see the promise to improve prediction, prevention and treatment of mental issues as bound to fail (Rose, 2018; Rosenberg, 2006; Timimi, 2014).

Even if one day we *could* draw on genomic information to accurately predict, prevent and treat psychiatric conditions, the normative question whether we *should* do so in the first place would still remain. The discussion about psychiatric diagnoses has indeed shown that in psychiatry opinions range from those of the medical community, who look for biological correlates of behavioural traits that it casts as 'deviant', to those of scholars who think that no biological entity underpins the 'myth' of mental conditions, which result from sociocultural values and sociopolitical processes (Timimi et al., 2011). In between these poles, some question whether milder symptoms should fall under the scope of psychiatrists' expertise, and others celebrate the positive traits that come with mental illnesses. This means that the agenda of psychiatric genomics may not be shared by all stakeholders in psychiatry. Thus, psychiatric uncertainty is not only a 'scientific' but also 'personal' issue (in Han et al.'s taxonomy), deriving from a lack of consensus on whether and how genomic information about mental traits should be used.

[13] I agree that these arguments seem to apply to mental health settings, but as reiterated in different passages in this section, I contest the exceptionality of psychiatry in this debate, and I accept that many of the above concerns about validity and reliability may be relevant to other areas of medicine. Drawing a thorough comparison among different branches of medicine is beyond the scope of this chapter.

In conclusion of this section, it is important to reflect on the relevance of these considerations to the present investigation of the applicability of Rose and Novas's traditional notion to psychiatric settings. Although Rose (2007) has hinted at anti-psychiatry attitudes, he does not seem to have extensively engaged with the implications that these have on his account of biological citizenship. The above paragraphs have shown that in psychiatry there exists a profound disagreement around whether certain behaviours should be seen as normal or abnormal, or as healthy or pathological. Therefore, not only is the actionability of genetic variants limited in psychiatry (s2.3.1.1), I also argue that to act according to the obligation to maximizing health and minimizing illness is *per se* difficult in a context where it is unclear what 'health' even means. Thus, the discussion of psychiatric uncertainty has shown that Rose and Novas's assumptions around the linear relationship between genetic risk and action and the consensus over the distinction between health and illness are of difficult portability to the context of psychiatric genomics.

The next section focuses on the second feature that is central to the ethics of psychiatric genomics, and that Rose seems to have neglected, i.e. the stigma attached to psychiatric conditions. I will discuss the impact that biogenetic attributions have on psychiatric stigma, to further investigate the transferability of the traditional notion to mental health.

2.3.2. Psychiatric stigma

Stigma as a psychological, social and ethical phenomenon has fascinated social scientists since the publication of Erving Goffman's pioneering work in 1963 (Goffman, 1963). Individuals with mental illnesses are among the most stigmatized groups (Bag et al., 2006) and the negative consequences that stigma has on their quality of life and opportunities for help-seeking behaviours (Corrigan et al., 2014), access to healthcare (Thornicroft et al., 2007) and employment opportunities (Wong and Lieh-Mak, 2001), are now well documented, although to date most research has centred around severe conditions (e.g. schizophrenia). Given that the present work focuses on the ethics of

neuropsychiatric genomics, here I will only discuss *biogenetic* explanations among other factors triggering stigma towards the mentally ill.

Early studies on psychiatric stigma suggested that, differently from physical traits, lay people tended to consider individuals responsible for their mental illness because of their 'poor character' or 'lack of moral backbone' (Corrigan and Watson, 2004, p. 477). This evidence led to mental health literacy initiatives based on the 'attribution theory' (Weiner et al., 1988), which postulates that people attribute causes to behaviours. By teaching the public about biogenetic causes of psychiatric disorders, which are beyond individuals' control, such initiatives aimed to generate empathy towards the mentally ill, while decreasing blame, and so stigmatizing and discriminatory attitudes (Australian Government. Department of Health and Ageing, 2013).

More recent empirical research has shown that, although biogenetic explanations are more widely accepted by patients, mental health professionals and the publics, psychiatric stigma still thrives, primarily because it is a much more complicated phenomenon than once thought (Haslam, 2011; Lebowitz, 2019; Schomerus et al., 2012). The 'mixed-blessings model' has been developed (Haslam and Kvaale, 2015). This acknowledges that, while biogenetic attributions decrease perception of blame, they simultaneously increase the perceived dangerousness and unpredictability of people with psychiatric conditions, a desire for social distance from them and pessimism about their chances for recovery (see Angermeyer et al., 2011; Kvaale et al., 2013; Larkings and Brown, 2018). Scholars interpret these findings in light of the literature on essentialist thinking.

Essentialism is a claim about identity (Wachbroit, 2000), according to which there is an unobserved and immutable essence that underlies the apparent characteristics of certain entities and defines their nature, or that is shared by members of a certain social group, and therefore explains their behaviour (Dar-Nimrod and Heine, 2011). Thus, *genetic* essentialism is defined as '[t]he tendency to infer a person's characteristics and behaviours as based on their perceived genetic makeup' (Dar-

Nimrod and Heine, 2011, p. 801); genes are used as an 'essence placeholder' (Medin and Ortony, 1989) to identify commonalities and differences between groups.

Essentialism is in part explained as a psychological tendency that helps us categorize entities in homogeneous and distinct groups and informs inferences about the behaviour of each group (Haslam et al., 2000). Although essentialist thinking is not problematic *per se*, essences are generally conceived as natural, fixed, inherent and boundary-defining. Because essentialist thinking involves the endorsement of the naturalistic fallacy of attaching moral worth to natural differences (Haslam, 2011), this psychological tendency often leads to stereotyping or developing negative notions about groups (Dar-Nimrod and Heine, 2011). Members of a group, who are seen as 'of a certain type', are stereotyped as 'other', 'different', 'dangerous', or 'of a reduced moral value' due to their real or supposed essence.

Scholars who interpret the results of research on psychiatric stigma as essentialism argue that the increased endorsement of biogenetic attributions of psychiatric disorders has triggered genetic essentialist thinking in psychiatry. This means that biology and genetics are used as the placeholder for the essence and identity of the mentally ill. Consequently, biogenetic causes of psychiatric disorders portray people with mental disorders as categorically different, thus causing desire for social distance; and because mental illness is seen as genetic in origin, it is perceived as unchangeable, which causes prognostic pessimism. In turn, because essentialist thinking is associated with stereotypes, accepting biogenetic causes reinforces stereotypes associated with mental illness, i.e. perceived dangerousness and unpredictability (Haslam and Kvaale, 2015).

These considerations show that psychiatric stigma deserves a thorough investigation in discussions about the ethics of neuropsychiatric genomics. Not only could the agenda of psychiatric genomics, if understood as aiming at the eradication of mental illnesses through genomic knowledge, reveal stigmatizing assumptions about psychiatric disorders as conditions that we *should* get rid of. Its

focus on biogenetic underpinnings of psychiatric disorders could also reinforce essentialist thinking and so stereotypes about the mentally ill.

In conclusion of this section, it is important to reflect on what research on biogenetic attributions and stigma illustrates about the shortcomings intrinsic in the traditional notion of biological citizenship. Although Rose and Novas have argued that technological advances have made biology amenable of control, hence inspiring an ethics of active citizenship, empirical evidence has shown that, even with regard to complex conditions like mental disorders (Kong et al., 2017), endorsement of biogenetic explanations leads to increased prognostic pessimism. Thus, not only may genomic information cause a paralysis of action (see parents' choices about prenatal and perinatal care in s2.2.2.2); if misinterpreted, in psychiatry it could also give rise to fatalistic and self-fulfilling attitudes. This further confirms that in practice individuals' awareness of genetic risk competes with their beliefs, values, understandings of probabilities and capacity to cope with genomic information, hence challenging Rose and Novas's assumptions of a linear relationship between acquisition of knowledge about genetic risk and action (assumption 2, s2.2.3), and of an alignment among duties, choices, values and desires (assumption 3, s2.2.3).

In this section, I have discussed the various layers of uncertainty that characterize psychiatric conditions and the stigma attached to them, to illustrate a range of limitations that emerge when the traditional notion of biological citizenship is applied to psychiatric settings. In the remainder of this chapter, I will further develop my critical take on Rose and Novas's account by focusing on young people as 'genomic citizens in the making' (Sabatello and Appelbaum, 2016).

2.4. Young people as 'genomic citizens in the making'

Before introducing the concept of 'genomic citizens in the making', it is important to position this thesis within the literature on childhood participation. Although I cannot undertake a detailed review of this scholarship, this section outlines the framework underpinning this thesis's focus on

young people. Following the terminology that is prevalent in this research area, unless otherwise specified the term 'child' will refer to all individuals from birth and under 18 years of age (Alderson and Morrow, 2011), and will be used as a synonym of 'young people'.

2.4.1. The western dominant conception of childhood

Within western societies, two epistemological assumptions have dominated sociological accounts of childhood until the end of the last century, namely children's *vulnerability* and *incompetence* as social, moral and political agents (Morrow and Richards, 1996). Vulnerability is understood both as a physical condition, i.e. children are weak, passive and subject to abuse; and as a structural condition, given that children lack the political, social and economic power that adults have (Lansdown, 1994). As a consequence of their alleged vulnerability, children are seen as dependent on adults in various domains of life. Incompetence is the idea that children lack the capacity to make decisions, because of their 'limited reasoning and morality' (Alderson, 2007, p. 2276), or because they are 'deficient in both rational and moral agency' (Gheaus, 2015, p. 16). This extends to the assumption of their incapacity to make decisions about research participation and clinical treatments; and to provide a valuable, or 'valid and reliable' (Singh, 2016a, p. 76), contribution to broader debates about social, political, and moral issues.

As sociological and philosophical investigations have suggested, these assumptions are grounded on Piagetian theories of child development, which emerged during the mid-20th century. They also draw from Ariès's thesis (1962) that 'childhood' as a separate social group only emerged in the Modern Ages with the introduction of compulsory education; and from western philosophers' 'hegemony of adulthood' (Burroughs, 2012), i.e. the idea that adulthood is the final stage of development, in which children reach full rationality, autonomy and social, political and moral agency (see Schapiro, 1999). These theories have contributed to the dominant conception of children as 'victims' in need of protection (Jans, 2004; Singh, 2016a) and have resulted in a cultural reluctance to listen to young people's voices (Morrow and Richards, 1996).

However, since the 1990s a new sensitivity towards children and their worlds has emerged (Gopnik, 2009; James and Prout, 1997; Morrow, 1996). While early sociological work focused on childhood only as the age of 'becoming adults', scholars have become interested in studying childhood *as childhood* in its own right or in the 'here-and-now' (Morrow, 2011). According to many scholars this officially started with or culminated in the ratification of the 1989 UN Convention on the Rights of the Child (UNCRC), which emphasizes children's rights to participation as active members of their families, communities and societies. In particular, Article 12 states that 'States Parties shall assure to the child who is capable of forming his or her own views the right to express those views freely in all matters affecting the child' (United Nations, 1989).

Subsequently, academic research on children and their perspectives has surged, leading to the new field of childhood studies, which contributes to the theoretical framework of this thesis. The aim of this interdisciplinary scholarship is to move beyond the epistemological assumptions about children's vulnerability and incompetence that still survive in the western world, in order to take children's views seriously in the various domains of political, social and moral life.

This does not mean that children' social, moral and political agency is taken to be equivalent to that of adults (Gheaus, 2015; Morrow and Richards, 1996). Although today's children are increasingly encouraged to express their voices in 'all matters affecting [them]', they are still considered as one of the social groups that is in most need of care. These recent developments have made it evident that children play an 'ambivalent' or 'ambiguous' role in society (Jans, 2004), positioned between protection and increasing participation. Nonetheless, by situating the present research within this emerging framework I want to challenge the idea that vulnerability (and incompetence) constitute the 'master identity' of children (Christensen, 2000, p. 40). My argument is that they are neither *unique* nor *inherent* traits of young people.

The dominant conception of childhood has indeed portrayed young people as lacking those characteristics that supposedly they will acquire in adulthood, i.e. independence, autonomy,

rationality and moral, social and political agency. These resonate with the traits of the neoliberal citizen that is valued within the traditional notion of biological citizenship; i.e. those of the 'rational, calculating [individual] whose moral autonomy is measured by their capacity for "self-care"' (Brown, 2003, para. 7, point 3). This description certainly does not map well onto the condition of a child, but the dominant conception of childhood has concealed that adults are often very distant from the ideal condition of complete rationality, agency and autonomy (Burroughs, 2012).

As the above review of the criticism of the traditional notion has shown, adults are not always rational actors; they are constantly in the process of developing and improving as agents in various social, moral and political domains; and they depend on networks of personal, legal and social supports and pressures, which explains why various groups in society do not match the profile of the 'good' biological citizen (s2.2.3). In this sense, vulnerability and incompetence are not unique to minors, and full rationality, autonomy and agency should not be a requirement for people's contributions to be taken seriously.

In addition, empirical studies have shown that with adequate time, support and experience children can reach a *degree* of capacity to make medical decisions (Steinberg, 2013), or contribute to debates about, for instance, gen-ethics issues (Bernhardt et al., 2003; Iredale et al., 2006), that resembles that of adults' abilities. Indeed, minors do not constitute a homogenous group (Nuffield Council on Bioethics, 2015), but have 'evolving capacities' (United Nations, 1989). Some have the cognitive abilities and emotional maturity to make their own decisions (Manzini and Vears, 2018). Alderson and colleagues' research on the competence that some children with long-term conditions have to consent to medical treatment or research is enlightening in this respect (Alderson, 2007; Alderson et al., 2006).

Children also possess abilities that are *qualitatively* different (or different *in nature*) from those of adults, or traits that most adults are likely to have lost. Evidence from philosophy (Matthews, 1984), experimental psychology (Gopnik, 2009) and sociology (Alderson, 2007; Alderson et al., 2006)

suggests that children are curious, imaginative, fast learners and (at least the younger ones) less influenced by social conventions. These are important conditions for reasoning and decision-making, for raising questions about and addressing moral, political and societal issues, and so they 'qualify children for some degree of both rational and moral agency' (Gheaus, 2015, p. 8). This may explain examples of children's engagement with moral dilemmas in a different way to adults, and their formulation of creative solutions that do not merely parrot adults' perspectives (see Singh, 2013). Thus, if children's capacities are not assessed against adult criteria, vulnerability and incompetence may cease to be seen as inherent in childhood.

In summary, a genuine concern with children's 'ambiguous' or 'ambivalent' position (and their alleged vulnerability and incompetence) requires adults to engage with their views in a way that accommodates their different (from adults') vulnerabilities and (in)competencies. It is against this approach to childhood that this thesis highlights the importance of listening to the ethical concerns that children as 'genomic citizens in the making' express about current genomic advances.

2.4.2. Genomic citizens in the making

As explained in Chapter 1, technological and methodological progress has allowed the shift from the study of rare monogenic conditions to that of common complex disorders, including psychiatric conditions, adding new nuances to the gen-ethics debate. Many empirical studies have investigated young people's understanding of the heritability of monogenic conditions, and their views of relevant ethical issues (among the most recent systematic reviews on the topic see Godino et al., 2016; McGill et al., 2019). Likewise, adults' attitudes towards advances in psychiatric genomics have been explored (Laegsgaard et al., 2009; Wilde et al., 2010). However, few studies have investigated young people's understanding of the biology of common complex disorders and their moral attitudes towards emerging genomic technologies, and no systematic research has done so in the specific context of mental health.

Young people's contributions are often overlooked in bioethics, because of the assumptions about children's vulnerability and incompetence, which in bioethics translates into recurrent scepticism towards their capacity to reflect on complex ethical issues (Singh, 2016a). However, young people's exclusion from ethics debates about neuropsychiatric genomics is problematic because, due to the unprecedented effort in this field, today's children are likely to be confronted with ethical questions that were less (or not) relevant to older generations. Therefore, it is important to enable young people to articulate what they value as normatively important in discussions around psychiatric genomics, in order to inform the ethics agenda for the future of mental health.

This is even more the case because young people occupy a peculiar place as citizens of the genomic era, as captured by the concept of genomic citizens in the making. This concept was introduced by Sabatello and Appelbaum (2016) in a recent paper that describes the rise of 'genomic citizenship' and reflects on its significance for the younger generation. In a way that resonates with Rose and Novas's tradition,[14] Sabatello and Appelbaum see genomic citizenship as requiring citizens to become informed about genomics, engage in genetic testing and other self-surveillance practices, and ideally participate in shaping policy around genomic issues. They argue that adolescents are genomic citizens 'in the making' in the sense that, differently from adults, they are still developing as genomic citizens; yet, they are 'particularly poised to fulfil the role of informed and engaged genomic citizens' (Sabatello and Appelbaum, 2016, p. 295) compared with younger children, as a result of their different developmental stage. Because Sabatello and Appelbaum define adolescents as > 13 years, they seem to attribute an age-based connotation to the term 'in the making'. However, in light of the above considerations about children's capacities, I will use the concept of genomic citizens in the making more broadly to refer to any child 'who is capable of forming his or her own views' (United Nations, 1989) about genomic issues.

[14] Although Sabatello and Appelbaum do not explicitly refer to Rose and Novas's framework, their profile of the genomic citizen is consistent with that of the 'traditional' biological citizen who engages in risk-management calculations.

The literature on children's understanding of genetic and genomic concepts and attitudes towards gen-ethics issues suggests that young people constitute a peculiar group of genomic citizens – they are 'in the making' – because of reasons related to their genomic literacy and genomic consumerism. Genomic literacy refers to having sufficient knowledge of genomics to be capable to make informed decisions about one's own health and contribute meaningfully to societal debates about genomic science (McInerney, 2002). Those who were born in the 21st century are the first generation to be raised in the age of rapid advances in genetics/genomics and of the widespread use of the internet (Rew et al., 2010), and so they are more likely than the older generations to be exposed to genomic information in educational contexts or to retrieve it online (Sabatello and Appelbaum, 2016). This may impact the way they perceive genomic information (Melas et al., 2012), and their moral attitudes towards genomic technologies may significantly differ from those of adults (Sabatello and Appelbaum, 2016), as they may be more familiar with genetic concepts, or more able to access online genomic explanations.

Despite some promising evidence from the US (Rew et al., 2010; Sabatello et al., 2019), these observations do not imply that young people's genetic literacy is necessarily better than that of the older generations; increased exposure to genomic education in school or genomic information through the media could be a double-edged sword (Sabatello, 2017). In the US, many school educational materials and genetics curricula are of poor quality, often failing to include key concepts such as gene–environment interaction (Sabatello, 2017), and many teachers do not feel prepared to teach contemporary genetics issues (Sabatello et al., 2019). Similarly, A-Level and undergraduate students in the UK still underperform in their genomic knowledge (Gallop et al., 2017), and a recent systematic review showed that children from both the general population and at-risk families have a general understanding of inheritance, but they lack deeper knowledge of complex genetic concepts and specific genes/carrier rates (McGill et al., 2019). Because the internet is an important source of genomic and other health information for minors (Jain and Bickham,

2014), and media representations of genetics are often inaccurate (Gallop et al., 2017; Marcon et al., 2018), young people are more likely to be exposed to misleading information about genomics available online, e.g. news that may boost their expectations of the potential of genomic technologies.

Similar considerations apply to young people's genomic consumerism (Sabatello, 2017), i.e. their increasing involvement in genomic services and initiatives. Genetic testing for monogenic conditions and next-generation sequencing have become increasingly common among minors (Mand et al., 2013; Sabatello and Appelbaum, 2016), because of research initiatives that enrol children, e.g. the 100,000 Genomes Project in the UK. Moreover, the marketing strategy of some direct-to-consumer companies specifically targets young people, which may attract minors whose parents are opposed to testing (Sabatello and Appelbaum, 2016). Furthermore, adolescents seem to be interested in accessing genomic services or having agency over them, as many believe they should be involved in decisions about testing and research participation (Bernhardt et al., 2003), even when they would like some parental input in the decision (Geller et al., 2003; Szybowska et al., 2007). Finally, professional opportunities in genomics that were unavailable in the past (e.g. bioinformatics and genetic counselling) are likely to become more popular career options among the younger generations (Sabatello et al., 2019).

However, children's increasing contact with genomic services has not been followed by a corresponding development in measures to empower them to take responsibility for choices regarding their genomic makeup, and to protect them as genomic consumers (Sabatello and Appelbaum, 2016). As in other domains of healthcare, young people are not considered as 'conventional' decision-makers and are not allowed to consent to genetic testing in clinical or research settings (Sabatello, 2017; Sabatello and Appelbaum, 2015). Moreover, evidence shows that some genetic professionals feel ill-equipped to communicate with young people, provide developmentally appropriate care and address conflicts between children's and other family

members' needs (Duncan and Young, 2013; Young et al., 2019). While this confirms the importance of investigating young people's views as emerging genomic citizens, which may inform the consumer behaviours they will hold when they reach the age of majority (Gunter et al., 1998; Kolarova, 2011) or decide to reproduce (Iredale et al., 2006; Melas et al., 2012), it also shows that young people are embedded in a 'troubling consumerism/agency gap' (Sabatello, 2017, p. 27).

It is important to consider what Sabatello and Appelbaum's concept of genomic citizens in the making says about Rose and Novas's account of biological citizenship. My argument is that children's limited genomic knowledge and agency mean that the traditional notion does not track well onto the condition of young people. In other words, it seems unlikely that children will be able to live up to the expectations imposed on the informed and rational genomic citizen, who should take responsibility over their genomic makeup to maximize their health. Thus, young people add to those groups whose position in society casts doubts on the third assumption undergirding Rose and Novas's account, which in s2.2.3 I called 'conformity among duties, choices and values'.

The above discussion about the notion of genomic citizens in the making has once again highlighted the 'ambiguous' or 'ambivalent' condition of young people (Jans, 2004), who oscillate between genomic literacy and vulnerability to genomic misinformation, and between growing consumerism and limited agency. However, in light of the dominant conception of childhood in the western world, some concluding remarks are necessary to ensure that the notion is not used to protect adults' hegemony in gen-ethics debates, rather than as a way to empower children.

As Burroughs has noted, because children are seen as deficient in comparison to adults, they are 'most often encountered in discussion of education and moral and political development' (2012, p. 2). For example, children's specific sensitivity about themes of global relevance (e.g. peace) is often used for educational purposes, or considered as a base for future rather than actual citizenship (Jans, 2004). Perhaps unintendedly, Sabatello and Appelbaum's concept of genomic citizens in the making falls in this tradition. Expressions like 'in the making' (Sabatello and Appelbaum, 2016, p.

295) or ‘raising’ genomic citizens (Sabatello and Appelbaum, 2016, p. 292), as well as the age-based connotation that the authors seem to attribute to the concept, suggest a perspective that focuses more on what children will *become* than on children of the genomic age in the *here-and-now*. The same applies to papers on young people’s understanding of genetics and attitudes towards gen-ethics, which often discuss implications for *formal education*, or strategies to better *inform* children about genomics issues (Bradbury et al., 2012; Gunter et al., 1998). Indeed, Sabatello and Appelbaum themselves argue: ‘Whether adolescents will succeed in this goal [i.e. to progressively assume the responsibilities of genomic citizens] depends on how well they are *nurtured* in the genomic era (emphasis added)’ (2016, p. 303).

I do not aim to undermine the importance of such an educational intent. However, the focus on children’s ‘becoming’ seems to be grounded on the idea that the traditional biological citizen is and should be the ultimate end goal of young people’s development, while this chapter has shown that there are reasons to be critical of the traditional framework of biological citizenship. Thus, the label ‘in the making’ should be used as a way to separate the position of young people from that of the traditional biological (or genomic) citizen, more than in the developmental sense that seems to be attributed to it by Sabatello and Appelbaum. Moreover, this top-down educational approach runs the risk of turning young people’s involvement in gen-ethics discussions into a missed opportunity for investigating what we can *learn* from listening to their views and attitudes. This is an equally important intent, given that today’s children are the generation that will have to deal with the consequences of current advances for the longest. Therefore, precisely because of children’s peculiar position as genomic citizens ‘in the making’, I argue that it is important to shift away from a narrow focus on education to investigate the actual contribution that they can provide to debates about current pressing issues in psychiatric genomics. That young people do not fit the profile of the traditional biological citizen further confirms that we should empirically investigate their views

and attitudes in discussions about the ethics of psychiatric genomics, and assess whether the underpinning values align with the moral framework of biological citizenship projects.

2.5. Concluding remarks: Putting the 'traditional notion' to the test

In this chapter I have critically reviewed Rose and Novas's traditional notion of biological citizenship to argue that it is grounded on three assumptions that need further investigation. These are: (1) an alleged consensus over the distinction between health and illness; (2) a linear relationship between genetic risk and action; and (3) a conformity among duties, choices and values. I have shown that two features in mental health, i.e. the uncertainties over the ontology, aetiology and diagnosis of psychiatric conditions and the stigma attached to them, challenge such assumptions. Thus, it is important to build on Rose and Novas's reflections on biological citizenship to conduct a systematic study that takes the peculiarities of the field of mental health seriously.

This literature review has also highlighted that young people have not been involved in discussions about the application of genomic science to mental health. This is an important gap in the literature because children are among those who will be most affected by current research on psychiatric genomics, and yet evidence shows that, as genomic citizens in the making, they occupy a peculiar position in society. Young people do not seem to match the profile of the traditional biological citizen, and so it is important to investigate the moral framework within which they operate when discussing pressing issues surrounding recent advances in psychiatric genomics.

By pulling these arguments together, in conclusion of this chapter I argue that the traditional notion of biological citizenship should be put to the test in relation to both *psychiatric settings* and *young people*. This requires that young people's knowledge and views of psychiatric genomics and their moral concerns about advances in psychiatric genomics are investigated to understand what being a genomic citizen in mental health means to them. While Sabatello (2017) has anticipated the

importance of including children in psychiatric genomic conversations, the significance of the present research is that it does so *empirically*.

An empirically grounded notion of biological citizenship in mental health settings from children's perspective allows young people to contribute to shape the ethics agenda for the future of practices at the intersection between genomics and mental health. This is particularly topical because children have joined a wider wave of youth activism that covers topics as diverse as climate change, gun violence and the right to an education (The Lancet, 2019), by leading an increasing number of awareness and de-stigmatizing campaigns around mental health in the UK (Booth, 2017) and on the global scale (Booysen, 2019). This suggests that the younger generation's attitudes towards psychiatric conditions may be different from older generations' views, and so young people should be given the space to shape the kind of sensitivity towards mental health that they hope to see in future societies. In the next chapters, I will present and discuss the empirical bioethics study that I have undertaken to this purpose, starting from the presentation of autism genomics as a case study.

Chapter 3. Methodology: Autism genomics as a case study

3.1. A case study approach and the choice of autism

Having made the case for why it is important to conduct a systematic investigation of young people's understanding of biological citizenship and genetic responsibility in psychiatric settings, in this chapter I expand on the rationale for choosing autism genomics as a case study. Given the interdisciplinary nature of this research three sets of literature are critically reviewed: the science literature on autism and autism genomics; the literature on autism studies; and the empirical literature on stakeholders' perspectives around advances in autism genomics.

'Case study' as a methodological tool could take various forms (Crowe et al., 2011; Platt, 2007). In this thesis, I adopted a case-study approach in the sense that I tested the notion of biological citizenship in mental health settings by engaging young people in ethics discussions around neuropsychiatric genomics, and I deemed it necessary to focus such discussions on one psychiatric condition only, rather than on a range of disorders. This was so as not to overwhelm the study participants who may have not had much knowledge of mental health. The choice of autism, among other psychiatric conditions, seemed to fit this research well because, as mentioned in Chapter 1 and as I will expand on in the next sections, various stakeholders have demonstrated significant interest in the genetics of autism since the 1970s. Moreover, autism is characterized by the various layers of complexity that, as I argued in s2.3, emerge from the application of genomic science to mental health (Figure 4). This means that an investigation of the meaning of biological citizenship in mental health settings that focuses on autism genomics fits the theoretical framework developed in Chapter 2.

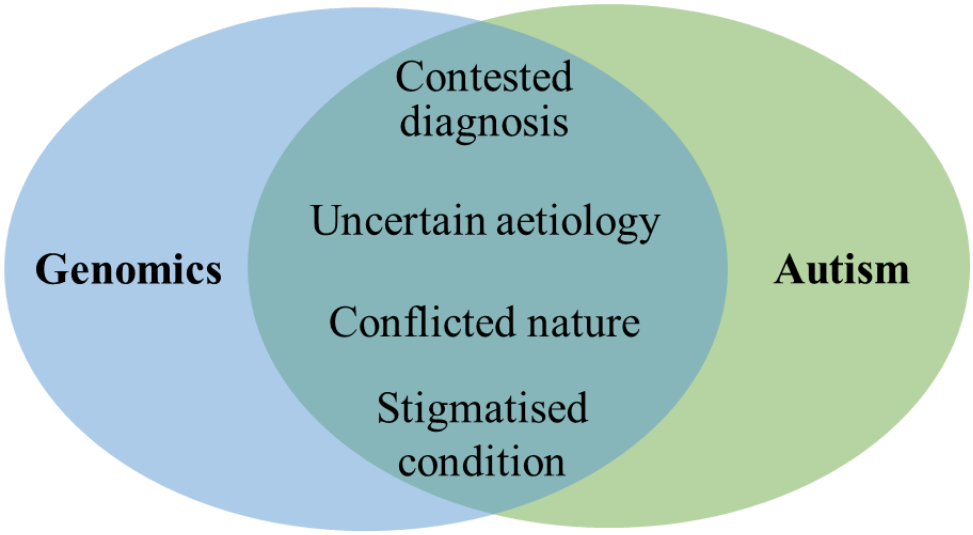


Figure 4: The various layers of complexity in autism

In the first part of this chapter, I will demonstrate that the diagnosis of autism, particularly its increasingly broad boundaries, has been contested (s3.2). I will then show that ASD aetiology is still largely unknown (s3.3) and that, at least in public debates, strongly polarized perspectives exist on the ontology and nature of autism, i.e. on what autism is and whether it should be conceptualized as a disorder, a disability, or a difference (s3.4). Throughout these sections it will become clear that stigma has been a driving force for stakeholders to promote certain understandings of autism and to push for certain research priorities, which have in turn informed the evolution of ASD diagnostic criteria.

Having showed that autism makes a good case study within an investigation of the meaning of biological citizenship in mental health settings, in the second part I will use the above-illustrated complexities as a background to review the empirical studies that have investigated stakeholders' perspectives about advances in autism genomics (s3.5). This review will allow to identify the main gaps in the existing empirical debate, which will be pulled together with the arguments made in

this chapter to further demonstrate the appropriateness of autism as a case study to test the traditional notion from the perspective of the general population of young people.

3.2. ASD contested diagnosis: A brief history of its evolution

The first investigation of 'autism' in psychiatry is credited to Leo Kanner, who published an article on 11 case studies in 1943 (Kanner, 1943), although 'infantile autism' officially entered the DSM in 1980, under the subclass of 'pervasive developmental disorders' (Silverman 2017). The specific diagnostic criteria for autism have evolved over time. The DSM-5 calls the condition 'autism spectrum disorder' (ASD) and defines it as a neurodevelopmental disorder generally diagnosed in early childhood (American Psychiatric Association, 2013). The diagnosis of ASD is behavioural and based on the observation of two main features, which are 'persistent deficits in social communication and social interaction across multiple contexts' and 'restricted and repetitive patterns of behaviour, interests, or activities' (American Psychiatric Association, 2013, p. 50). Similar criteria are used in the World Health Organization's *International Classification of Diseases, 11th edition* or ICD-11 (2018).

Social communication difficulties may constrain a child's learning in their first few years, while repetitive behaviours tend to 'interfere with eating and sleeping and make routine care (e.g. haircuts, dental work) extremely difficult' (American Psychiatric Association, 2013, p. 57). In adulthood, gaining independence may be difficult for some autistics. Most autistic people also experience hyper- or hyposensitivity to certain stimuli (e.g. sounds, light, or texture), which may cause intense distress and trigger extreme behavioural responses. Moreover, people with ASD tend to have comorbidities (e.g. epilepsy, sleep problems and gastrointestinal issues), as well as psychiatric conditions, such as anxiety and depression (especially in adolescents and adults), and specific learning difficulties (American Psychiatric Association, 2013). Many metabolic conditions have also been associated with ASD (Schaefer et al., 2013; Wang et al., 2015).

Both the DSM-5 and the ICD-11 recognize the heterogeneous clinical presentation of autism. The idea that autism is a spectrum disorder was first developed in the 1970s (Silverman, 2017; Wing and Gould, 1979), but it was only with the DSM-5 that, by merging several DSM-IV subcategories, the clinical diagnosis of 'autism spectrum disorder' was introduced. The autism spectrum includes, on the one end, about 75% of individuals with mild to severe intellectual disabilities; many have language impairments with 30% of them being minimally verbal, and they generally require care and support throughout their life (Baker and Jeste, 2015). On the other end of the spectrum, autism includes individuals who are conventionally, but not without controversy (Alvares et al., 2019), considered to have 'high-functioning autism',[15] i.e. they have 'normal' or 'above normal' IQ (Baron-Cohen, 2002). They tend to live independent lives, and some may be highly skilled in tasks that match their specific interests (American Psychiatric Association, 2013). Those people with average to above-average IQ without language disabilities are said to have Asperger syndrome (Asperger, 1991), which is among the subcategories that were merged into the ASD diagnosis in the DSM-5. Given this clinical heterogeneity, the DSM-5 recommends that diagnosis includes a description of the severity of presentation of the individual's symptoms (mild, moderate, or severe).

The new diagnostic criteria and terminology are supposed to better account for the idea that autistic traits manifest across a spectrum (Baker and Jeste, 2015), which aligns with the increasing recognition of the dimensionality of psychiatric conditions (s2.3.1.1). They are also meant to address the poor validity and reliability of the DSM-IV diagnostic subtypes (Hazen et al., 2013). However, these and other changes in the DSM-5 have been contested by scholars and advocacy groups (Hazen et al., 2013; Pickersgill, 2014), not least because of the widening of ASD diagnostic criteria, which started in the late 1980s (Singh, 2016b) and has led to a diagnosis that now

[15] Like other autism-related terms, 'high-functioning', 'low-functioning', 'mild' and 'severe' autism are contested. When used in this thesis, they should not be interpreted as lack of awareness of the existing debate.

encompasses traits from severe impairments to mild atypical neurodevelopmental differences (Graf et al., 2017).

The broadening of diagnostic boundaries, together with sociocultural factors such as the increased awareness about autistic difficulties, contribute to explain the steep increase in the prevalence of ASD in the recent years, which according to the latest official estimates accounts for one out of 100 adults in the UK (Brugha et al., 2012). Although the reliability of this and similar prevalence rates have been questioned because of the inconsistent use of methodologies across studies (Fombonne, 2018), there seems to be clear evidence that autism is four times more common in boys than girls (Fombonne, 2003; Schaefer, 2016), which may be due to genetic factors (the 'female protective effect', see Robinson et al., 2013), or diagnostic biases (Baker and Jeste, 2015).

Moreover, it is because of the heterogeneous traits included in such a broad diagnosis, which seem to fall under the remit of different clinical specialties, that '[a]utism has been treated alternatively as a psychological, neurological, behavioural, or genetic disorder' (Silverman, 2017, p. 32). Two among the most-known theories that have for long influenced the understanding of autism are the so-called 'refrigerator mother' theory and the causal role of childhood vaccines in ASD development. The first gained attention in the 1960s, particularly in Bettelheim's articulation (1967), according to which children raised by cold and hard-hearted families withdraw from the social world to find comfort in their loneliness, thereby showing autistic symptoms. The second was formulated in the 1990s, when Wakefield and his colleagues claimed to have identified evidence of a link between the measles, mumps, and rubella (MMR) vaccine and ASD development (Silverman, 2017).

Many psychiatrists have rejected Bettelheim's arguments, which have drawn blame and suffering on a large number of autistic people's parents (Silverman, 2017); and in 2010 *The Lancet* retracted Wakefield and colleagues' article, because of evidence of their research misconduct (The Editors of The Lancet, 2010). That vaccines do not cause autism has been demonstrated in several

subsequent studies and meta-analyses (see Taylor et al., 2014). Nonetheless, therapies based on psychoanalytic theories (e.g. 'packing therapy') are still used in France, the UK and the US, despite having been criticized as unethical, cruel and ineffective (Amaral et al., 2011; Boris and Zeanah, 2005). Moreover, across the western world there has been a recent increase in anti-vaccination movements, in part driven by fear of a link between vaccines and autism (Hussain et al., 2018).

Among autism scholars, it is widely accepted that the survival of these theories, particularly psychoanalytic ones that had fuelled a parent-blaming culture, led to an increased orientation towards biological explanations, i.e. 'external' causes of autism (Bumiller, 2009; Fitzgerald, 2012; Silverman, 2017; Singh, 2016b) – a hypothesis that fits the discussion about psychiatric stigma, blame and biogenetic attributions (s2.3.2). The 'geneticization of autism' (Navon and Eyal, 2014), which found early confirmation in the first twin studies in the 1970s (Folstein and Rutter, 1977), was appealing to parents because it offered hope for the development of effective treatments and, most importantly, exculpation from the responsibility of causing their children's difficulties. Some scholars have argued that the families of autistic children played a crucial role in allowing the rise of the epistemic infrastructure for autism to 'become' a genetic condition (Silverman, 2017; Singh, 2016b). By the end of the 1990s autism was indeed designated as 'one of the psychiatric disorders most influenced by genetic factors' (Fombonne, 1998, p. 349).

However, in the early 2000s it became clear that there is no 'gene for' autism (Singh, 2016b), hence creating a conflict among scientists. Some see recent advances in the understanding of the biology of ASD as leading to more questions than answers; others consider the knowledge acquired as a source of optimism for future discoveries. The former are supported by those social scientists who understand the geneticization of autism as the result of cultural and societal processes initiated by various stakeholders (Grinker, 2008; Nadesan, 2005); for the latter, there is strong evidence that ASD is a genetic condition (Schaefer, 2016), although so far ASD genomic research has only allowed the identification of a clear genetic aetiology in a small number of cases.

3.3. Recent advances in and uncertainties about autism genomics

Since the late 1970s twin studies have been conducted to investigate the genetic heritability of autism. Concordance rate in monozygotic twins (MZ) is currently calculated as between 36% and 95%, and between 0% and 31% in dizygotic twins (DZ) (Schaefer, 2016). Studies on population genetics have shown that heritability estimates are around 0.65–0.8 (Pettersson et al., 2018), and couples' risk of having subsequent children with autism ('recurrence risk') is 10–20% if they already have one autistic child, and 30–50% if more than one (Schaefer, 2016). Given the 4:1 ratio of males to females mentioned above, the recurrence risk is higher for male siblings of autistic people (Brandler and Sebat, 2015).

Twin studies have been used to support the theory that autism is a genetic condition; however, their methods have been highly criticized for being biased or based on unsupported assumptions (Joseph, 2004, 2006, cited in Timimi et al., 2011). For instance, concordance rates in MZ twins have not taken into account differences in twins' autistic traits, which may significantly differ due to non-shared environmental factors (Castelbaum et al., 2019). Moreover, the above estimates indicate that genetics does not account for the entirety of autism risk; although specific environmental and epigenetic mechanisms are still under investigation, evidence suggests that some environmental exposures, such as advanced parental age, complications during pregnancy or birth, extreme preterm birth and *in utero* exposure to certain air polluters increase autism risk (Ciernia and LaSalle, 2016; Muhle et al., 2018).

However, that autism has a genetic component has been confirmed by the identification of specific genetic risk factors since the introduction of microarray and, subsequently, next-generation sequencing techniques and genome-wide association studies. This made it possible to shift away from failed attempts at discovering a 'gene for' autism, and embrace a genomic framework to understand the condition (Singh, 2016b). Although this is a rapidly evolving field, existing

evidence suggests that there are three genomic groups among individuals diagnosed with ASD, which can be divided into syndromic and non-syndromic (or idiopathic) cases (Box 4).

Box 4: Three genomic groups of ASD cases

Syndromic cases

In 10% of cases of autism, mutations in a single gene cause some syndromes (e.g. Fragile X or Rett syndrome) that in turn cause autistic traits. People with syndromic autism are more likely to have intellectual disabilities or a higher degree of cognitive impairment (Wang et al., 2015). However, only one-third of people with Fragile X show autistic traits, which present differently than in people whose primary diagnosis is ASD (Thurman et al., 2014), and children with Rett syndrome should not be automatically diagnosed with ASD, because their social interaction difficulties tend to decrease when they grow up (American Psychiatric Association, 2013). Thus, some have questioned whether these should be considered 'classic' cases of autism or primarily as genetic conditions that may cause autism-like symptoms (Wright, 2014).

Non-syndromic (or idiopathic) cases

Copy number variations

Chromosomal microarray analysis (CMA) can identify copy number variations (or CNVs, see Box 3 above for a definition) in 15–20% of cases of neurodevelopmental disorders, including autism (Miller et al., 2010a), although estimates vary depending on the study cohort (Tammimies et al., 2015). CNVs are high-confidence variants, but taken individually they have a limited effect on the risk of the development of autism, and because they tend to involve larger strands of the DNA, pinpointing the specific factor responsible for a specific ASD trait has been difficult so far (Wang et al., 2015). Moreover, determining the clinical utility of CNVs is challenging. Indeed, some CNVs associated with autism have been found in control samples, e.g. family members who do not have autistic symptoms (D'Abate et al., 2019; Weiss et al., 2008); in people who show some symptoms that fall under the ASD spectrum (e.g. speech and language delay), but do not make up for an ASD diagnosis (Hanson et al., 2010; Myers et al., 2020); and in individuals with other psychiatric conditions (Niarchou et al., 2019). CNVs associated with autism have also been identified in genetic pathways of medical conditions (e.g. certain cancers), which may require immediate or long-term care (Markkanen et al., 2016).

Single-nucleotide polymorphisms

For ~70% individuals currently diagnosed with autism identifying a specific genetic aetiology has proved to be difficult (Schaefer, 2016). These cases are most likely caused by single-nucleotide polymorphisms (SNPs, for a definition see Box 3), i.e. common variants with small effect size that act in concert and within epigenetic processes and mechanisms of gene–gene and gene–environment interaction (Muhle et al., 2018). The contribution of SNPs in the development of ASD is still being explored to enable derivation of polygenic risk scores.

In summary, because ASD susceptibility genes have complex inheritance, incomplete penetrance and variable expressivity, the aetiology of autism is still uncertain. Its environmental contributions

are not well understood, and recent studies have discovered hundreds of genetic variants associated with autism (Coe et al., 2019), which may lead to different degrees of severity of ASD. At present we do not seem to have enough knowledge to correlate genomic information to specific degrees of severity of an autistic person's difficulties, except for syndromic cases, which are associated with a higher degree of cognitive impairment and epilepsy and a greater risk of co-morbidity (Baker and Jeste, 2015; Folstein and Rosen-Sheidley, 2001; Schaefer, 2016). Furthermore, the same mutations that increase people's risk of developing autism may predispose them to a range of other psychiatric and medical problems (Carroll and Owen, 2009; Markkanen et al., 2016). Thus, in Han et al.'s (2011) taxonomy (s2.3.1), the uncertainty around the aetiology of autism derives from the probabilistic, ambiguous and complex nature of ASD genomic information.

This is why I sympathize with Timimi and colleagues, who have argued: 'What is disappointing is the continued instance of many researchers and writers in this field that biological causation for autism (as we define it today) is already a given' (2011, p. 138). In fact, the genetic heterogeneity that characterizes such biological causation, coupled with the phenotypic heterogeneity discussed in the previous section, explain why few majorly relevant results of genetic and genomic research have been translated into clinical applications, as I discuss below.

3.3.1. Rationale and challenges of using genomic information about autism

Currently, genetic testing is primarily used as a confirmatory diagnostic tool on children who have received a diagnosis of autism based on a behavioural assessment. To provide a molecular diagnosis, combining various types of genetic testing is sometimes necessary (Box 5).

Box 5: Clinical genetic evaluation of children diagnosed with ASD (Wang et al., 2015)
• In cases of unexplained autism or when a syndromic case is highly suspected, genetic testing for single-gene syndromes (e.g. Fragile X in males) is recommended (Schaefer, 2016).

- In idiopathic cases, CMA to detect CNVs has so far been considered as a first-tier genetic test (Miller et al., 2010a), i.e. estimates of its diagnostic yield[16] have been the highest compared with other tests. However, recent evidence suggests that next-generation sequencing technologies (NGS), particularly exome sequencing, should be used as first-tier diagnostic tests in place of CMA. NGS allows the identification of variants that CMA cannot detect (Srivastava et al., 2019), but may also lead to the identification of VUS (for a definition see s2.2.2.2) and incidental findings, which provide information about DNA changes 'unrelated to the primary purpose for testing' (National Library of Medicine (US). Genetics Home Reference, 2020c).
- In both clinical and research settings, tests are often conducted 'in trios' to compare the DNA of a child with their parents' DNA and assess whether the identified variants are inherited or *de novo* (Wang et al., 2015). Some diagnostic laboratories also offer ASD targeted gene panels, which include genes associated with syndromic and non-syndromic ASD.
- At the moment no other medical testing is routinely recommended for ASD (Baker and Jeste, 2015). For instance, although increasing research has been devoted to the exploration of autistic people's atypical brain development, 'there is no evidence to support routine neuroimaging in autistic individuals' (Muhle et al., 2018, p. 520).

It is worth noting that this field presents regulatory inconsistencies. In the UK, genetic testing is not recommended by the National Institute for Health and Care Excellence (NICE) guidelines (2017) unless autism is comorbid with developmental delay or other significant features suggestive of a syndrome. Across Europe, different laws have been implemented and, in the absence of an internationally approved list of genes with strong clinical ties to autism (Wright, 2019), even within the same country different laboratories offer different testing or test different variants (Zarasca, 2019a). In contrast, the American College of Medical Genetics and Genomics (ACMG) 2013 practice guidelines recommend genetic testing on all children with a confirmed diagnosis of ASD (Schaefer et al., 2013).

Although evidence shows that only a minority of families are actually offered testing (Kiely et al., 2016), the rationale underpinning the ACMG recommendations is that genetic testing could provide a 'definitive' or 'unifying' diagnosis to the child and their family (Schaefer et al., 2013, p. 400).

[16] Although ASD diagnosis is still behavioural, the scientific literature uses 'diagnostic yield' to indicate the proportion of autistic people who carry a known pathogenic CNV over the total number of individuals tested.

Because of the complexity of autism genomics, only about 25% of autistic individuals are confirmed as having an identifiable genetic cause for their condition (International Society of Psychiatric Genetics, 2019), but in empirical studies parents have reported important psychological benefits from receiving a molecular diagnosis of their child's autism, as I will discuss in section 3.5.

It is important to note that advances in the discovery of genomic indicators of autism raise hopes and promises that are not confined to a more 'objective' diagnosis, but extend to improved 'primary prevention', i.e. the complete prevention of autism prenatally, and 'secondary prevention', which consists in reducing the impact of ASD, or predicting autism and intervening as early as possible to slow its progress or lessen its risk. Three sets of interventions that propose to use the knowledge of autism genomics for the prediction, prevention and treatment of ASD at different life stages could be developed or are under development: (1) prenatal diagnosis (PND) and preimplantation genetic diagnosis (PGD); (2) genetic tests performed on infants for early intervention purposes; and (3) pharmacological treatments (Figure 5).

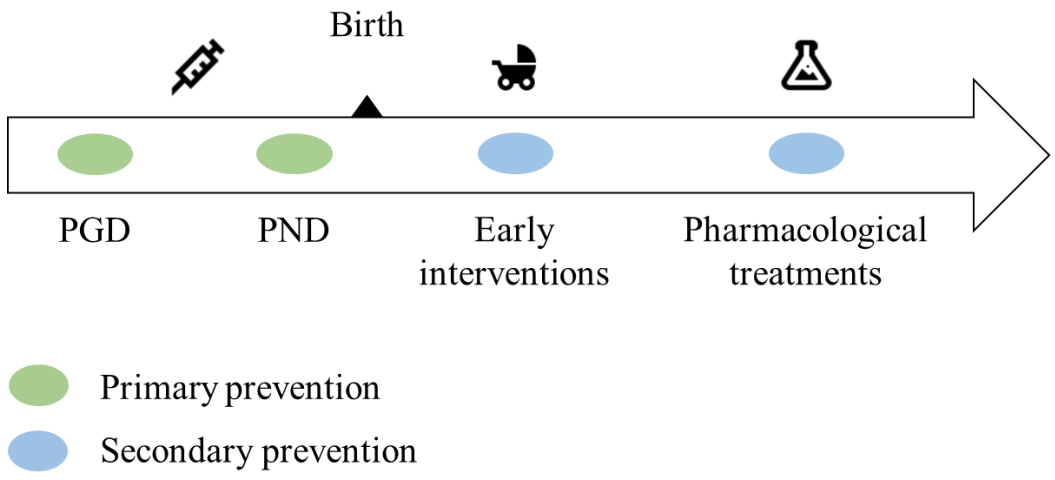


Figure 5: The primary and secondary prevention of autism

In the next pages, I will briefly discuss these interventions, on which the empirical part of this research focused (see Chapter 4).

3.3.1.1. Primary prevention

Preimplantation genetic diagnosis and prenatal diagnosis, introduced in Chapter 2, are not standard applications of genomic science in the context of autism, because most variants (especially CNVs) associated with autism are *de novo*, and so identifying at-risk pregnancies is difficult. Moreover, genomic information collected prenatally about autism has little clinical validity, except for single-gene mutations responsible for syndromic cases or for a few highly penetrant CNVs.

Clear guidelines on the prenatal use of ASD genomic information still need to be developed. At the moment, PGD is offered to couples at risk of transferring 'serious inherited diseases' or genetic and chromosomal conditions 'where a specific gene is known to cause that condition' (Human Fertilisation and Embryology Authority, 2019), i.e. Fragile X or Rett syndrome in the context of autism. Interestingly (and controversially), in 2012–2013 the Western Australian Reproductive Technology Council listed autism among the 'genetic conditions' approved for PGD (2013, p. 13), and allowed at-risk couples to receive PGD for sex selection to reduce the risk of having an autistic child (Amor and Cameron, 2008; O'Leary, 2013), given the 4:1 ratio of males to females.

During pregnancy, screening can identify increased risk of certain foetal abnormalities (e.g. kidney abnormalities) associated with microdeletions and microduplications that are also linked to autism, although this is an indication of risk that needs to be confirmed by diagnostic testing. The latest guidelines by the American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine (2016) recommend prenatal CMA to detect CNVs only for patients whose ultrasound has identified one or more major structural abnormalities, although CMA is increasingly used for other indications, e.g. advanced maternal age (Bernhardt et al., 2012). In the UK, only highly penetrant CNVs are reported prenatally when foetal anomalies detected on screening or familial risk have triggered gene testing for such variants (Cuthbert, 2018). NGS is instead

recommended only in research settings (American College of Obstetricians and Gynecologists and Society for Maternal-Fetal Medicine, 2016), but it may become more common as costs decrease (Zarasca, 2019b), raising ethical concerns around incidental findings and VUS.

Scholars have indeed argued that increasing pressure from high-risk families may soon lead to more widespread use of information about autism genomics for reproductive decision-making (Hoang et al., 2018a; Tammimies et al., 2015; Wang et al., 2015). In the meantime, a few commercial laboratories, such as GeneDx in the US, already offer prenatal testing for known familial pathogenic variants in at-risk pregnancies (GeneDx, 2018).

3.3.1.2. Secondary prevention

Early interventions for autism

The early intervention (EI) paradigm, which has become a priority in the context of autism (McGuire, 2013), argues that the earlier a child is diagnosed with autism, the earlier they will receive interventions to address their symptoms and the better their outcomes will be. Since the early 2000s, the number of prospective studies to identify early behavioural or developmental signs of autism has increased, because of evidence of the long time lag between parents' first concerns about their children's development and their children's official diagnosis (Szatmari et al., 2016). Many of these studies use high-risk[17] infant sibling design (or HR design), because younger siblings of autistic people ('baby sibs' or 'infant siblings') have a familial susceptibility to it, and among those who do not receive an ASD diagnosis, 30–40% show subclinical autistic traits and atypical development (Charman et al., 2017). Social-communicative and sensory-regulatory deficits (e.g. failure to respond to one's own name by 12 months, avoiding eye contact, having delayed speech, or hyper-responsiveness to stimuli) are now considered as early behavioural symptoms of autism (Centers for Disease Control and Prevention, 2019a).

[17] In section 3.5.2, I will explain that some stakeholders oppose the use of the 'risk' language to describe younger siblings of autistic children.

When a child shows early ASD behaviours or is diagnosed with autism, enrolling them in early intervention programmes aims to limit the impact that autism will have on their development. A range of strategies fall under the broad category of EIs, and some are particularly intensive in terms of time and financial commitment (Russell, 2016). Some scholars list behavioural and educational interventions only, such as the well-established yet contested applied behavioural analysis (ABA, see s3.4.2); early intensive behavioural intervention (EIBI), which uses the principles of ABA; speech and language therapy; and social skills instruction. Others also include family support, and medical management of comorbid health issues, such as seizures and gastrointestinal problems, which as aforementioned are prevalent in autistic individuals (Myers et al., 2007).

Despite evidence that EIs based on early behavioural diagnosis lead to better developmental outcomes, EI studies have several limitations (Landa, 2018; Russell, 2016). Identifying reliable early behavioural signs of autism is challenging because currently used tools lack sensitivity and specificity (Bradshaw et al., 2015; Jones et al., 2014). It is often hard to distinguish between signs that should warrant further investigation and difficulties that are transient and will resolve in typical developmental trajectories (Walsh et al., 2011). Thus, although the American Academy of Paediatrics recommends autism-specific screening on all children starting at 18 months (Hyman et al., 2020) because of evidence that 'ASD can be diagnosed accurately before two years of age' (Zwaigenbaum et al., 2015a, p. S61), the median age of ASD diagnosis in children in the US seems to be much higher (Centers for Disease Control and Prevention, MMWR 2012). Seemingly, in the UK the median age (55 months) did not decrease between 2004 and 2014 (Brett et al., 2016).

Moreover, most EI strategies have not been tested in randomized controlled trials (RCTs), and so it is difficult to pinpoint the specific component that makes them effective. For instance, in a recent systematic review, only one in five studies on the effectiveness of EIBI was RCT, and the authors rated the overall quality of evidence as low or very low (Reichow et al., 2018). Furthermore, some scholars are concerned about issues of generalizability of behavioural signs (and effectiveness of

interventions) that are identified in HR design studies (Szatmari et al., 2016), or in studies that are conducted predominantly with white children (Monnier, 2018).

This is why scientists hope that a combination of biological markers (i.e. genetic variants and neural differences) associated with ASD will inform improved early identification of emerging cases of autism (Geddes, 2015; Walsh et al., 2011) and more effective EI strategies. At the moment, no genetic marker is recommended for early ASD screening (Zwaigenbaum et al., 2015b), because their clinical utility still needs to be demonstrated. However, at least within the infant sibling population the detection of ASD-relevant CNVs seems to predict ASD or atypical development, especially if coupled with polygenic risk scores and brain imaging; this may help inform risk estimates for 'intensified surveillance' of baby sibs' behaviour (D'Abate et al., 2019, para. 14). Moreover, genetic testing could one day identify individuals who might benefit from EIs targeted to subgroups of at-risk people with a similar genotype.

Some pre-symptomatic tests are already commercially available. For instance, notwithstanding intense criticisms (Jordan and Tsai, 2010), the ARISk Test was launched by the biotech company IntegraGen to identify ASD-associated SNPs in infants from multiplex families as early as six months and categorize them as at low, medium, or very high risk of autism (IntegraGen, 2012).

Pharmacological treatments for autism

At the moment there is no 'cure' for core autistic traits (Loth et al., 2016),[18] i.e. social communication difficulties and repetitive behaviours. Pharmacological treatments should only be used to ameliorate associated features for which there exist approved medications in children, such as depression, anxiety, sleep problems, gastrointestinal issues and epilepsy (NHS, 2020). Based on existing evidence, NICE guidelines (2013) only recommend antipsychotic medication for what is named 'behaviour that challenges', i.e. behaviours that might cause harm to others or the autistic

[18] Nor are 'cures' necessarily a hoped-for outcome of the identification of ASD biomarkers, as will become clearer below.

person (e.g. head banging and hand and wrist biting). Because they do not have UK marketing authorization for use in children in this way, antipsychotics should only be used when psychosocial or other interventions have been unsuccessful.

In 2014, antipsychotics to address aggressive behaviour or irritability, particularly Risperidone, were the most widely prescribed drugs for ASD across Europe, Asia, Oceania, Central America, South America and Africa (Wong et al., 2014). Together with the antipsychotic Aripiprazole, Risperidone is the only FDA-approved pharmacological treatment for ASD (Ghosh et al., 2013). However, a recent study has shown that in the UK a much wider range of psychotropic medications are used off-label for autistic people, including children, and that use increases with age and psychiatric comorbidities, although rates are still lower than in the US (Houghton et al., 2018).

Antipsychotics have significant limitations when used for autism, as for other psychiatric diagnoses (Rose, 2018). They seem to work only with a small proportion of autistic individuals, which may be due to the heterogeneity of patients' clinical phenotype, and when they are effective they cause serious side effects, e.g. weight gain, sedation and tachycardia (Baker and Jeste, 2015). Thus, NICE recommends clinicians to 'review the effectiveness and any side effects of the medication after three to four weeks', and to 'stop treatment if there is no indication of a clinically important response at six weeks' (2013, p. 14), especially in light of the little knowledge of their long-term effects on children.

Failure to identify effective treatments for core ASD traits and associated symptoms, and parents' frustration at being unable to support their autistic children, explains why some parents have resorted to alternative biomedical interventions (Nadesan, 2005; Silverman, 2017). These are exclusion (e.g. gluten-free) diets, vitamin supplements, chelation therapies and hormone therapies, which are advertised as offering miracle and rapid solutions that other interventions fail to provide, but are discouraged and described as potentially harmful in official reports of conventional medicine (Centers for Disease Control and Prevention, 2019b; NHS, 2020; NICE, 2013).

Lack of effective treatments may also explain continued research efforts to unfold the biological mechanisms underpinning autism (Silverman, 2017). Studies on single-gene disorders linked to autism have been assumed as paving the way for rapid advances in the development of more effective medications for ASD (Baker and Jeste, 2015; Ghosh et al., 2013). High hopes were generated in the late 1990s by the identification of the mutations responsible for Rett syndrome (Guy et al., 2007), but they were later disappointed when research failed to identify biological and epigenetic processes to develop specific therapies for affected children, and derive investigative techniques that could be used in ASD research (Silverman, 2017).

Nonetheless, some scientists and members of the autism community are still hopeful that genetic testing may inform more individualized diagnosis and treatment. Because some CNVs are linked to medical issues that warrant either immediate or long-term monitoring (e.g. epilepsy or susceptibility to certain cancers), molecular diagnosis can already inform treatment options (Tammimies et al., 2015; Walsh et al., 2011). One day, advances in autism genomics may allow improved stratification of autistic people in pathophysiologically relevant diagnostic groups, i.e. subgroups of people whose autism is the result of the same biological pathway, and who may respond well to certain behavioural therapies or drugs with the same treatment target (Loth et al., 2016). The first pharmacological treatments based on genotype for core symptoms of autism are already being tested on subgroups of autistics (ClinicalTrials.gov, 2018; Muhle et al., 2018). In turn, patient stratification may improve trial design thanks to a more rational selection of participants, thereby increasing the chances for progress in the identification of effective treatments (Ghosh et al., 2013). Finally, gene editing to repair specific genetic loci playing a role in ASD development is often included among the possible future applications of autism treatments (Chung, 2019), although uncertainties about the potential long-term effects mean that it is not currently being explored beyond animal models and syndromic forms of autism (Muhle et al., 2018).

3.3.2. From a descriptive to a normative matter

Although the three sets of interventions illustrated in Figure 5 above are not yet part of standard care, some scientists have predicted that findings in autism genomics will be translated into clinical applications in the next five to 10 years (Muhle et al., 2018). However, close scrutiny should be exercised on scientists' 'promissory' discourse (Smart and Martin, 2006) around the potential for autism genomics to lead to better prediction, prevention and treatment of autism. The results of genomic studies have shown much more uncertainty than once expected, which has led critics to question whether there is any coherent biological entity underlying the broad diagnosis of ASD (Bovell, 2015; Timimi et al., 2011). Reflecting on the persistent uncertainties about the diagnosis and aetiology of the condition, already in the early 2000s Nadesan had highlighted that the problem in autism studies resides in assuming that 'there *are* distinct autistic phenotypes and genotypes that can be *reliably* identified and measured across populations' (2005, pp. 139).

I argue that even if technological and methodological improvements could one day lead to the 'reliable' identification of 'distinct autistic phenotypes and genotypes', there would still remain a more critical question to answer, i.e. whether autism should be predicted, prevented and treated in the first place. No matter how much our understanding of the genomic underpinnings of autism progresses, autism genomics will not provide a solution to this fundamental question, because this is not only a 'scientific' issue, but also a 'personal' one (see s2.3.1, Han et al., 2011; Newson et al., 2016). In other words, deciding whether certain behaviours are 'normal' (if such a thing exists at all), and what to do about 'abnormal' ones, requires an exercise of balancing scientific knowledge with moral values and societal constructions. The next section will show how this exercise has caused profound disagreements in 'expert' and 'public' debates around autism.

3.4. ASD conflicted nature: Disorder, disability, or difference?

The uncertainties about the diagnosis and aetiology of autism are reflected in the variety of stakeholders' conceptualizations of the 'ontology' of autism (what autism *is*), and of its nature, i.e. the value judgements attached to such ontology. The multiplicity of meanings that are included under the label of autism has caused divisions and disagreements, to the point that any account of the various voices that are part of the autistic and autism community[19] is inevitably incomplete.

In this section, I distinguish among three models of autism. The medical model understands autism as a disorder to be predicted, prevented or 'cured'; the social model considers autism as a disability that results from society's lack of commitment to accommodating autistic people's needs; and the neurodiversity model describes autism as a valuable part of human variation that should be celebrated. I show that to some extent this is an artificial distinction because the three models are not mutually exclusive. Thus, I conclude by arguing that, although at times the public debate is still highly polarized, the prevailing understanding of autism has now shifted to a multifaceted account that tries to hold these different conceptualizations together.

3.4.1. The medical and social models of autism

The medical and social models were introduced in Chapter 2; here I discuss them in the context of autism, where they express two different stances regarding the search for an ASD cure. The former adopts an 'essentialist understanding' of autism (Baker, 2011), according to which ASD is a *disorder* causing challenges that are solely the result of autistic traits; in turn, these are reduced to biological mechanisms within the individual. In other words, the medical model embraces the idea that autism consists of a range of biological dysfunctions or deficits – in fact, until the early 2000s

[19] In this chapter I will adopt the distinction, drawn by autism scholars such as Orsini (2009), between the 'autism movement', which refers to activists and advocates concerned with pressing for policy change around autism treatments and interested in autism causes; and the 'autistic movement', which refers to activists who use, although not exclusively, a disability rights framework to create a positive identity around autism.

it downplayed the role of other complex factors in the development of autism. Such model finds expression in the DSM's interpretation of behaviours, and promotes a 'cure-oriented agenda' (Baker, 2011) that seeks biomedical knowledge about the deficits experienced by autistic people to ultimately prevent, control and (at least in its critics' view) eliminate them (Kapp et al., 2013).

The success of this model is reflected in the current pattern of research expenditures on autism. According to the latest evidence, across the world private and government funds prioritize studies on basic sciences, including neuroscience, cognitive systems and genetics, which have received increasing funding in the last couple of decades (Interagency Autism Coordinating Committee, 2016; Pellicano et al., 2013; Singh et al., 2009). Research in these areas has been boosted by non-autistic parents who have endorsed the medical model to counteract stigmatizing theories that blamed them for the development of their child's autism (s2.3.2). In perfect alignment with the profile of the responsible and enterprising 'traditional' biological citizen, in the early 1990s, and especially in the US, these parents have founded a range of organizations with a shared interest in accelerating and funding biomedical research on autism to develop effective pharmacological treatments that could lead to cure or recovery, or that could help children behave as 'normally' as possible (Kapp et al., 2013; Singh et al., 2009). Jennifer Singh (2016) has provided a comprehensive account of some of these organizations, particularly the National Alliance for Autism Research and Cure Autism Now, which have merged into the charity Autism Speaks, an avid (and controversially so) supporter of research to develop ASD treatments.

In contrast, the social model of autism, which emerged from the broader social model of disability, has provided the autistic movement with the framework to build a positive identity around autism (Orsini, 2009). Applying the social model theory to autism means acknowledging that autistics experience some difficulties; however, these are understood as the consequence of a *disability* caused by societal structures that disadvantage people on the spectrum (Kapp et al., 2013). The social model adopts a 'constructivist understanding' of autism (Baker, 2011), which in its most

radical form considers society as uniquely responsible for autistic challenges, while in its moderate version sees autistic challenges as the result of the interaction between ASD traits and contextual factors. It supports a 'cause-oriented agenda' that advocates for autistic people's civil rights to promote a philosophy of social acceptance, barrier removal, equal opportunities and inclusion (Baker, 2011; Jaarsma and Welin, 2012). While the medical model, with its heightened focus on the smallest components of organisms, identifies in the individual the *locus* for intervention, the social model shifts the onus of change from the individual to society (Russell et al., 2017).

It is thanks to these efforts for recognition of structural disadvantages on autistic people's lives, as well as to the higher involvement in priority-setting demanded by autistics and their allies, that the resource prioritization pattern described above has recently seen a proportional decrease in the number of studies on basic sciences and an increase in other areas relevant to the lives of autistic people (Singh et al., 2009). Some autism experts have even spoken of 'a new era for autism research' (Pellicano et al., 2018). For instance, although basic sciences remain a priority, the 2016–2017 (and latest) strategic plan of the US Interagency Autism Coordinating Committee (IACC), which advises the Secretary of Health and Human Services on issues related to autism, has shifted towards improving services meant to 'maximize quality of life, and minimize disability, while also ensuring that individuals on the autism spectrum are accepted, included and integrated in all aspects of community life' (Interagency Autism Coordinating Committee, 2017, p. VI). Some of the claims of the social model have inspired the activism of neurodiversity advocates.

3.4.2. The neurodiversity model of autism

The term 'neurodiversity' was first used in the late 1990s by the sociologist and Asperger activist Judy Singer (Ortega, 2009). It refers to the spectrum of variation in neurocognitive functioning that characterizes both the brains of 'neurodivergent' people, e.g. people with autism, attention deficit hyperactivity disorder, or dyslexia, and those of neurotypicals. The neurodiversity model, and the movement that promotes it, rejects the stigmatizing connotation that the medical model attaches to

autism and other forms of neurodivergence, which it does not see as pathological but as *differences* that are part of the spectrum of human variation, resulting from the different way autistics' brains are 'wired' compared to neurotypicals' (Ortega and Choudhury, 2011). According to this view, autism is an inseparable and intrinsic component of autistics' identity (Jaarsma and Welin, 2012; Kapp et al., 2013; Walsh, 2010). In parallel to the Deaf culture (s2.2.2.2), some scholars have even talked about the emergence of an autistic culture (Ortega, 2009).

The neurodiversity movement emerged when (adult) autistic self-advocates, including those who struggle to socialize or are non-verbal, found in the spread of the internet the means to promote a positive narrative around autism (Brownlow and O'Dell, 2013; Nadesan, 2005; Orsini, 2009). Thus, the biological fact of diversity has become the basis for demanding the societal recognition, acceptance, and inclusion of neurodivergent people initiated by the social model, from which the neurodiversity movement originated (Orsini, 2009). Some neurodiversity advocates have moved beyond the 'cause-oriented' agenda of the social model to align with a 'celebration-oriented agenda' (Baker, 2011) that considers autistic identity as a gift to be proud of (Ortega, 2013).

In contrast to the strong version of the social model, most neurodiversity activists do not see autistic difficulties as emerging from oppressive societal structures only, but promote a rise in 'neurological self-awareness' (Singer, 1999, p. 65), i.e. they put a strong emphasis on the idea that autism is natural, innate and ultimately reduced to biological and genetic mechanisms happening within people's brains. Thus, they favour brain-centred approaches to the understanding of personhood, and neurological and neuroscientific terms and metaphors to describe the self, although these remain somewhat vague in advocates' writings, given that to date no specific difference has been identified in autistics' neurological makeup (Ortega, 2009; Ortega, 2013).

This way of conceptualizing autism creates a paradoxical situation in which the heterogeneity of human brains is used to support acceptance and celebration of diversity; yet, autistic people's brains are made homogenous and distinct from those of non-autistics, who constitute a homogeneous (but separate) group (Ortega, 2009). This is at odds with the idea of the autism spectrum (Ortega and

Choudhury, 2011) and may reinforce essentialist and potentially stigmatizing assumptions, according to which autistic people are 'of a different kind' (Nadesan, 2005). However, it serves the purpose of creating a shared identity among autistic people to respond to those parents of children with severe symptoms, who argue that the neurodiversity movement only speaks to the interests of 'high-functioning' autistics (Ortega, 2009; Ortega, 2013).

Moreover, attributing autism to a different brain functioning liberates autistic people from the blame attached to their 'odd' behaviours (Ortega, 2013). This allows a dialogue between neurodiversity advocates and parents who push for advances in the biomedical knowledge of autism to counteract stigmatizing theories about their parenting styles (Kapp, 2020). However, the neurodiversity movement has been criticized, both internally and by those who hold anti-psychiatry sentiments, for creating a reductionist and solipsistic identity politics around autism that too readily embraces neurological explanations of human behaviour that are typical of the medical model (Zifendorf, 2005).

The above paragraphs suggest that there are overlaps between the medical and neurodiversity models. As Ne'eman, founder and former president of the Autistic Self Advocacy Network, explained: 'We agree that research and the scientific method is the best way to ascertain what autism is. We agree that autism […] has its roots in the brain and is predominantly genetics in origin' (2019). Indeed, both paradigms use a 'brain language' and adopt an essentialist rather than constructivist understanding of autistic traits, and most self-advocates support the use of DSM diagnostic categories to grant access to essential services (Kapp, 2020). However, the two models radically disagree about the consequences of such an understanding of autism (Ne'eman, 2019; Silverman, 2017), given that self-advocates interpret the cure-oriented agenda of the medical model as aspiring to complete normalization or eradication of autistics' identity, grounded on ideas that express negative judgements towards autistic people (Baker, 2011; Orsini, 2009).

This is why ABA, currently one of the most popular autism behavioural therapies, which was developed to make autistic children 'indistinguishable from their normal friends' (Lovaas, 1987, p. 8), has caused tensions between parents and autistic self-advocates. The latter accuse ABA therapists of aiming to normalize autistic behaviours and using (at times painful) punishments as behavioural modifications to achieve this goal (Ne'eman, 2019), and claim that parents who support such interventions should not be the primary spokespersons for their autistic children (Orsini, 2009).

Seemingly, inspired by the claims of the social model, neurodiversity advocates have raised concerns about the fact that genomic research has 'crowded out the areas of research that are of greatest priority for autistic people and our families' (Ne'eman, 2019). This does not mean that they reject genomic research completely, but only when aimed at the prevention and 'cure' of autism. Nor does this mean that neurodiversity advocates oppose any form of support for people on the spectrum. The neurodiversity movement acknowledges that autism can be disabling at times (Davidson and Orsini, 2013); thus, one of its core mandates is to ensure that individuals on the spectrum can access relevant services (Ne'eman, 2010). However, therapies and support should target symptoms that directly impair the individual and their quality of life, rather than harmless traits such as non-injurious 'stimming', i.e. repetitive body movements that, being noticeable, some parents consider as source of social stigma, but for many autistics serve as coping mechanisms in situations that cause intense emotions (Kapp et al., 2013; Kapp et al., 2019).

3.4.3. Beyond polarizations: Finding a common ground among autism models

As Davidson and Orsini have pointed out, the public debate about the nature of autism, and how and whether autism should be treated, is polarized 'especially in online forums dedicated to autism research and advocacy' (2013, p. 2). However, it is important to note that in 'real life' many autistic people do not exclusively cling to one pole of the debate rather than the other. On the contrary, Ortega has used the expression 'cohabitation of everyday ontologies' (2013) to argue that autistics

oscillate among different identities, at times more psychological and at others more neurobiological, to refer to themselves and others.

There is evidence that autistics who support the neurodiversity movement make efforts to appear more neurotypical, not as an end goal, but as a pragmatic strategy to cope with certain situations, so as not to be confronted with stigmatizing attitudes that can be more limiting than autistic difficulties *per se* (Jones and Meldal, 2001). Moreover, self-identification as autistic and awareness of neurodiversity is not only compatible with acknowledgement of autistic difficulties and support for interventions to ameliorate them (Kapp et al., 2013); in a study on autistic adults' views of their own strengths, participants presented some of their traits as advantageous or disadvantageous depending on the social context, the extent to which they manifest them, their capacity to control them and the perspective from which their behaviours are judged (Russell et al., 2019).

Further, for some in the autistic and autism community, particularly those with severe difficulties and their family members, biomedical research aiming at developing ASD treatments is important, if not a priority (Concrick, 2013; Russell et al., 2017). Prominent autistic activists have publicly expressed their support for medications (Grandin, 2006), and some have accused the social model rhetoric of being representative of 'high-functioning' autistics only (Mitchell, 2008; Rubin, 2005). Conversely, not all parents of autistic people are in favour of the development of cures for ASD (Ortega, 2009), despite being aware of the hurdles that having autism or caring for an autistic child can cause (Russell et al., 2017). Moreover, autism scholars have argued that proponents of a pure medical model who deny any role to social structures in the emergence of autistic difficulties 'are virtually impossible to locate' (Baker, 2011, p. 10).

Therefore, following developments in the broader field of disability studies (s2.2.2.1), scholars' prevailing understanding of autism is shifting to a multifaceted account that tries to find a common ground among the different models (Hens, 2019). These efforts have culminated in the rise of the interdisciplinary field of critical autism studies, where 'critical' refers to the rejection of any

reductive representations of autism (Davidson and Orsini, 2013; O'Dell et al., 2016). The uncertainties that, as I showed, characterize autism mean that a radical polarization of views is unsustainable and does not serve the interests of autistic people (Davidson and Orsini, 2013). On the contrary, an account that converges different models and adopts multiple agendas offers the opportunity of accommodating the varied experiences of people on the spectrum and reduces the chances of trivializing the struggles that some of them might encounter.

Because the neurodiversity movement adopts claims from both the medical and the social models, neurodiversity scholars have argued that an interpretation of their movement based on the biopsychosocial approach of functioning, which recognizes that biological, psychological and social factors can contribute to or interfere with autistics' adaptive functioning (Engel, 1977; World Health Organization, 2001), offers the opportunity for an integrative model of ASD (Kapp, 2013). Those who adopt this approach accept that different interventions may be appropriate for different people across the spectrum to achieve good quality of life, including medications for some, while this does not exclude that a positive identity can be constructed around ASD (Kapp, 2013; Ne'eman, 2019). Kapp et al. have called this the 'deficit-as-difference' model of autism, which transcends 'a false dichotomy between celebrating differences and ameliorating deficits' (2013, p. 66). In this way, the neurodiversity movement makes a discourse that depathologizes autism, while allowing space for amelioration of the difficulties linked to it (Nadesan, 2005).

The present chapter has so far discussed the various layers of uncertainty characterizing the study of autism, which concern the diagnosis, aetiology and ontology or nature of autism; as shown, discussions around these uncertainties have been influenced by stigmatizing attitudes toward ASD. This demonstrates that autism serves as a good case study for research that aims at testing the traditional notion of biological citizenship in light of the complexities that become evident when genomic science is applied to mental health. The next section presents the results of a literature review of the empirical studies that have investigated autism stakeholders' perspectives around the

use of genomic information about autism, to further expand on the rationale for the choice of autism in this research.

3.5. The socio-empirical debate on autism genomics

In the past decade several empirical studies, most of which were conducted in the US, have investigated stakeholders' views and concerns around advances in autism genomics. Appendix A provides an overview of the papers identified in this body of literature. These include both qualitative and quantitative research primarily conducted with relatives of autistic people, the majority of whom are parents of autistic children. Some papers engaged professionals whose main expertise is autism, including researchers, psychologists, psychiatrists, educational specialists and other practitioners in health settings; a minority of studies involved autistic adults among other participants.

Most empirical literature focuses on parents' attitudes towards clinical genetic testing for ASD and professionals and parents' views on autism genomic research, including issues related to providing and receiving individual research results. This is not surprising because the aetiology of ASD is still uncertain and so research on the genomic underpinnings of autism is abundant, and genetic testing as a confirmatory tool for a behavioural diagnosis is the only translational application of genomics currently recommended in routine care in the context of autism. Other areas covered by this literature include prenatal genetic testing for autism; genetic testing to assess the risk for ASD; pharmaceutical interventions for autism; priority-setting in autism research; and the sub-field of research on early identification of and intervention on ASD ('early autism research').

Similar findings were identified across studies, and they confirmed many of the ethical concerns discussed throughout this chapter. In this section, I briefly discuss these findings under the categories of autism genetic testing and autism research to highlight the gaps in the existing debate.

3.5.1. Autism genetic testing

The empirical studies on genetic testing for autism primarily investigate parents' awareness, experiences, attitudes and information needs relative to clinical tests (i.e. CMA). In only two studies had some participants direct experience of testing (Narcisa et al., 2013; Reiff et al., 2015), while in the others parents had never heard of or had never been offered testing. This confirms that, despite existing guidelines, awareness and uptake of CMA is low among parents of autistic children (Li et al., 2016; Xu et al., 2016).

Both parents who had their children tested and those confronted with a hypothetical scenario were in favour of CMA. One of the features most strongly associated with perceptions of usefulness of CMA or willingness to test was the opportunity for receiving a causal explanation of a child's ASD, which could in turn offer peace of mind, networking opportunities with families with a similar genotype and relief from feelings of guilt (Johannessen et al., 2017; Reiff et al., 2015; Xu et al., 2016). No participant raised the argument, discussed by autism scholars (Singh, 2016b), that both inherited variants and *de novo* CNVs, which most likely emerge in gametes from environmental exposures, may implicate parents in new forms of responsibility for their child's ASD.

A molecular diagnosis offered some parents a renewed understanding of ASD as a 'concrete' condition, and so greater acceptance of their child's behaviours (Reiff et al., 2015). This, I argue, might reveal genetic essentialist assumptions. Interestingly, autistic adults have also expressed essentialist views albeit in a different way, i.e. by raising the concern that a negative test result could disprove the ASD diagnosis they had integrated in their identity (Singh, 2016b). Parents also mentioned the opportunity for starting early behavioural interventions, ruling out serious genetic disorders such as DiGeorge syndrome (Johannessen et al., 2017; Reiff et al., 2015; Singh, 2016b; Xu et al., 2016) and accessing further educational opportunities upon learning that children with a certain variant can develop reading skills (Reiff et al., 2015). Many participants hoped that genetic testing could advance knowledge about the biological underpinnings of ASD for the development

of effective behavioural or pharmaceutical treatments (Xu and Floyd, 2017; Xu et al., 2016). However, in one study on professionals' views of ASD genetic testing and counselling, participants remarked that many of these benefits would only apply to a small subset of children who carry a known genetic variant with a known action plan and prognosis (Hens et al., 2016b).

Some respondents valued the opportunity for better understanding recurrence risk, not necessarily to inform reproductive decisions, but to be prepared for the possibility of another autistic child (Xu et al., 2016). However, scientists have highlighted that calculating the recurrence risk may be easier in syndromic cases or idiopathic cases caused by highly penetrant mutations than in those caused by 'weak' genetic factors, for which geneticists rely on familial risk (Hoang et al., 2018b).

Being prepared for the future also motivated parents' positive responses in the only two studies investigating their attitudes towards testing to assess the risk for ASD in their younger undiagnosed children (Narcisa et al., 2013), and towards prenatal genetic testing (PGT) for autism (Chen et al., 2015). In the latter study from the US, arguments in favour of termination of pregnancy following PGT were primarily proposed by Asian participants because of stigmatizing attitudes toward ASD in their community, while some parents argued they would continue their pregnancy despite a positive test result because of the bond with or acceptance of their existing autistic children. ASD professionals considered PGT acceptable only if additional known factors could reduce the future child's quality of life, or could make it too hard for parents to raise another autistic child (Hens et al., 2016b).

Despite the overall enthusiasm, issues around reduced clinical utility of genetic testing were raised by parents who had received negative results or VUS. In some cases, a child's behavioural diagnosis had already led to some interventions that were not affected after testing, or parents explained that their children's personality and behavioural traits were more informative than any available genomic information (Reiff et al., 2015). Moreover, positive results could cause psychosocial harm, e.g. blaming one side of the family for passing on pathogenic variants,

increasing anxiety about a child's future (Johannessen et al., 2017), or labelling a child at a young age (Narcisa et al., 2013). Finally, parents faced with a hypothetical testing scenario raised many practical considerations that would influence their decision, including costs, logistics of testing, negative impact on health insurance (Johannessen et al., 2017; Xu et al., 2016), accuracy of tests (Li et al., 2016) and the specific procedures involved (Li et al., 2016; Xu et al., 2016).

3.5.2. Autism research

Studies that fall under this category have focused primarily on parents' experiences and expectations of participating in ASD genomic research, which in most cases requires the collection of genetic material from both parents and their autistic children; and their perspectives about receiving research results. All studies reported parents' positive attitudes towards autism genomic research. Among those who have had direct experience, many enrolled in research to act altruistically (Tabor et al., 2011; Trottier et al., 2013). In Singh's study (2014), 'diagnostic parents' understood participation as an opportunity to receive a free diagnosis as part of the research study; and 'obligated parents', also described by Tabor et al. (2011), enrolled in research as part of their broader effort to acquire more knowledge about autism to help their autistic child in any possible way. Thus, Singh argues that parents exercise a form of biological citizenship in which normative choices about research participation do not rotate around traditional and abstract bioethical principles (e.g. autonomy, privacy and confidentiality), but are made within families' structural constraints and social complexities (e.g. their need for early diagnosis and access to services).

Some parents with no experience of research considered contribution to the development of better treatments among the main reasons for why they would decide to enrol (Johannessen et al., 2016). Other factors that parents would take or have taken into consideration when making such decisions included confidentiality of research results and the extent to which participants would be kept informed about the progress of research (Johannessen et al., 2016). Some also mentioned practical considerations about protecting children from unnecessary harms, research logistics, potential

rewards from research participation (e.g. access to better ASD interventions), the study relevance to the family's priorities, and its perceived importance to society (Tabor et al., 2011).

Most papers investigating parents' perspectives around receiving their children's research results presented participants with hypothetical scenarios, because the majority of genomic studies they were involved in did not allow return of results. Once again parents expressed strongly positive attitudes. These were grounded on practical interests that resonate with those discussed in the above section, i.e. predicting other health issues linked to certain variants and so personalizing ASD interventions (Trottier et al., 2013); informing reproductive decisions (Baret and Godard, 2011); and increasing societal acceptance of autism as a 'real' biological condition to leverage additional state support and decrease the publics' negative judgements about autistic behaviours (Trottier et al., 2013). Among parents who participated in research only to better understand autism and its causes and contribute to advance knowledge (Baret and Godard, 2011), some would want research results if available, but they would nonetheless derive satisfaction and a general feeling of hope from being proactive, taking control over the uncertainties surrounding their children's autism, and connecting with ASD experts and other families (Trottier et al., 2013).

In some cases, parents' positive attitudes meant that they did not perceive any potential harm in receiving individual autism genomic research results (Baret and Godard, 2011). When explicitly asked, some participants were optimistic about the power of future research to resolve uncertain results (Trottier et al., 2013). However, in other studies parents were sceptical about the long-term impact of non-medical benefits that could be derived from disclosure of results (Miller et al., 2010b), and some worried about the psychological implications of genomic information retrieved by research, which, similarly to considerations about clinical genetic testing, included feelings of blame, guilt, anxiety and stress (Tabor et al., 2011).

A minority of studies investigated autism professionals' attitudes towards ASD genomic research (Hayeems et al., 2011; Hens et al., 2016c; Miller et al., 2010b). Autism researchers raised concerns

about the clinical validity of research results (Hayeems et al., 2011). Miller et al. (2010b) noted that the greater endorsement of the genetic causation of autism that some participants expressed correlated with increased support for returning findings. Because of their uncertainty about whether autism is a 'disease' or a 'difference', in Hens et al.'s research (2016b) psychologists, clinical geneticists, educational specialists, paediatricians, child neurologists and child psychiatrists questioned some potential outcomes of ASD genomic research. These include the desirability of medications as a way to normalize autistic people, and of prenatal screening, which some professionals saw as in opposition to the positive narrative around autism that they aimed to develop with their patients with milder forms of autism.

Negative attitudes towards ASD medications were also held by a diverse group of stakeholders, including autistic adults, parents of autistic children and autism professionals, who were asked to comment on the research agenda of EU-AIMS, a large pan-European consortium studying the biological mechanisms of autism (Russell et al., 2017). However, respondents identified some conditions under which treatment should be considered acceptable, particularly if it helped manage stressful situations or co-occurring problems, rather than having autism as a generic target; and provided that medications did not become a convenient form of social control, but autistics could remain in control of whether, how and why to be treated. Of note, parents of severely autistic children were more in agreement with the idea that autism is a disease that can be harmful, and supported drug development for ASD, which highlights the importance of engaging with stakeholders with diverse experiences of and relations to autism.

In addition to Russell and colleagues' research (2017), the only studies that purposively recruited autistic adults, although among other participants, were those about research priority-setting (Frazier et al., 2018; Pellicano et al., 2014) and early autism research (Fletcher-Watson et al., 2017). Both the UK-based research by Pellicano et al. (2014) and the international research by Frazier et al. (2018) showed that participants were disappointed that current patterns of autism research are

biased towards basic sciences, and they stressed the need for future investments on issues of immediate practical concern to the autistic and autism community (e.g. better access to services and support). Differently from other groups, autistic adults did not support research on interventions targeting core symptoms of autism and meant to normalize people on the spectrum. In Frazier et al. (2018), a small subset of respondents, most of whom are autistic, opposed all autism-related research, or only approved research focused on co-occurring conditions, adult transition, lifespan issues and health and wellbeing.

Finally, only two studies investigated parents', autism professionals' and autistic adults' perspectives on early autism research (Fletcher-Watson et al., 2017; Fletcher-Watson et al., 2019). In contrast with the aforementioned uneasiness of some stakeholders with research on basic sciences, participants ranked genetic basis of ASD and early ASD signs in infants as the most important questions scientists should be asking about early autism. This might be because genetics is perceived as particularly relevant to research focused on very early life. However, autistic adults questioned early identification, better clinical knowledge of early signs and development of skills that prevent children from later receiving an ASD diagnosis as goals of this research area, while they favoured research to better understand the unique development of autism and its environmental contributions. This may be due to the different conceptualization of autism that some people on the spectrum have compared to other stakeholders. Except for healthcare professionals, all groups highlighted their discomfort with the 'at-risk' language used in early autism research to refer to baby siblings, and favoured expressions such as 'infants with high autism likelihood', or 'infants with higher chance of developing autism'.

3.5.3. Key themes and gaps in the socio-empirical literature

The above presentation of the empirical studies on stakeholders' perspectives on advances in ASD genomics suggests that some key themes can be identified in this body of literature. Parents and autism professionals seemed to hold positive attitudes towards the use of genomic information

about autism in clinical and research settings, in light of the acknowledgement of the medical and non-medical benefits that could be offered by clinical, prenatal and pre-symptomatic genetic testing for autism, and by research on the genomic underpinnings and early signs of autism.

However, concerns were also raised. Among these, parents tended to focus primarily on potential harms to their children and families, rather than making considerations about broader societal implications. Interestingly, in the few studies that involved autistic adults, their views tended to converge towards those of other stakeholders. However, by proposing arguments that recall the neurodiversity narrative, autistic people opposed the development of drugs for autism, early diagnosis and interventions to prevent a diagnosis later in life, which suggests that, if asked, they would hold similar attitudes towards prenatal tests. It is important to note, though, that these views may not be representative of all voices on the autism spectrum, as it is likely that autistic participants had milder difficulties (Fletcher-Watson et al., 2017). As I will explain in the next chapter (s4.3.2.1), to date there is no approved research method to elicit the views of autistic people with lower IQ and/or significant communication difficulties.

In conclusion of this section, it is important to highlight the gaps that emerge from this body of literature. Three key considerations are relevant to the discussions that will follow in this thesis. First, the above paragraphs have revealed that the empirical debate has so far involved autistic people, their family members and professionals invested in the study of autism. Less attention has been paid to the publics' attitudes towards such advances, and young people have been completely excluded from this debate. However, as argued in Chapter 1, technological advances are not only relevant to those they most directly affect, but also function as points of resonance for the way in which society thinks of a certain phenomenon. Listening to the publics' voices is, therefore, particularly important because autistics do not live in a vacuum but in societies with neurotypicals, whose attitudes towards advances in autism genomics are likely to both be informed by and influence the broader societal desire (or lack thereof) for the use of genomic information to predict,

prevent, or treat autism. Young people, in particular, should be engaged in these discussions because recent advances in autism genomics will require them to deal with ethical questions that were less (or not) relevant to older generations (s2.4.2).

Second, despite the extensive academic and public debate around the way in which autism should be conceptualized, and the value judgements attached to autism in the different models (s3.4), most empirical studies have failed to contextualize people's attitudes towards advances in ASD genomics within a broader investigation of their views on autism. Only one study linked researchers' perspectives on returning research results to their understanding of the aetiology of autism (Miller et al., 2010b), and only two papers referred to stakeholders' different conceptualizations of autism to account for differences in their support for medications and prenatal screening (Hens et al., 2016c), and for early ASD detection and intervention (Fletcher-Watson et al., 2017). In so doing, the empirical literature has missed the opportunity for shedding light on the values about and views of autism that underpin societal attitudes towards interventions for the prediction, prevention and treatment of ASD. To address this gap, in the empirical study that will be presented in the next chapters, the investigation of young people's attitudes towards the interventions followed a preliminary discussion about their understanding of what autism is, what it is caused by and whether they saw autism as a disorder, a disability, or a difference.

Finally, many social science scholars referenced in this chapter have drawn on empirical evidence to reflect on the concept of biological citizenship in the context of autism (Brownlow and O'Dell, 2013; Bumiller, 2009; Orsini, 2009; Ortega, 2009; Silverman, 2017). Moreover, the stakeholders' claims reviewed in this section support some of the considerations about biological citizenship that I have made so far in this thesis. Some parents, especially those who seek genomic information to inform reproductive decisions or support research for the prevention of autistic traits later in life, do seem to enact 'traditional' forms of genetic responsibility. For others, engagement with the ASD genomic apparatus is instrumental to achieve the pragmatic goal of alleviating some of the

difficulties that their families encounter, regardless of obtaining genomic information *per se*. This confirms that in genomic settings choices are not only based on awareness of genetic risk, but require the negotiation of a range of considerations, including situationally specific interests that Rose and Novas seem to have overlooked. Autistic adults and their allies seem instead to align with those citizens who, as described in Chapter 2, reject biotechnological advances, when these ultimately aim at preventing, curing, or eliminating their condition.

However, only one empirical study among those identified has discussed stakeholders' attitudes towards participation in autism genomic research within the tradition of biological citizenship (Singh, 2014). This was part of Jennifer Singh's broader sociological work (2016) on the different ways in which citizenship is exercised in relation to the social production of autism genomic knowledge. Pulled together with the first two considerations, this means that so far no systematic research has provided a picture of young people's views of autism and attitudes towards advances in autism genomics, in order to investigate the meaning that biological citizenship and genetic responsibility have for young people from the general population. This confirms the appropriateness of the choice of autism as a case study in the present research.

3.6. Concluding remarks: The rationale for this case study

Given the focus of the present research, any chosen case study needs to respond to the aim of investigating biological citizenship both in *mental health settings* and from the perspective of *young people*. In the first part of this chapter, I showed that autism is characterized by those complexities that emerge from the application of genomic science to mental health and that challenge Rose's choice to ultimately align citizenship in psychiatric settings to his broader conception of life in the biomedical age (s2.3). I demonstrated that the diagnosis of autism has been contested, not just because of its evolution over time, but also because of its increasingly broad criteria. Despite autism being considered as one of the most heritable psychiatric conditions, I showed that its aetiology is

still largely unclear, and I argued that advances in autism genomics will not resolve the disagreement over whether autism should be seen as a disorder to be predicted, prevented and treated, a disability to be accommodated, or a difference to be celebrated. Throughout these pages, I showed that debates around the ASD diagnosis, aetiology and nature have been influenced by existing stigmatizing attitudes towards autism.

In the second part, I reviewed the empirical papers that have investigated stakeholders' views of and attitudes towards advances in autism genomics, which have primarily focused on clinical genetic testing for autism and autism genomic research. I showed that the population involved, particularly parents and autism professionals, hold positive attitudes towards the use of ASD genomic information, while autistic adults have reported concerns over interventions to cure ASD or intervene early in its development. I identified three gaps in the existing literature, concerning the lack of involvement of the publics, and of young people in particular; the limited investigation of stakeholders' views of autism (i.e. its traits, aetiology and nature) that underpin their attitudes towards advances in ASD genomics; and the limited discussion of these attitudes through the lenses of the scholarship on biological citizenship. In this way, I have justified the choice of autism to test the traditional notion from the perspective of the general population of young people, and I have anticipated some of the methodological choices on which I will further expand in the next chapter.

Chapter 4. Methodology: Theoretical issues and methods design

4.1. Recap of research questions, aims and objectives

In the preceding chapters, I introduced the theoretical framework of this research, situated at the intersection between the literature on biological citizenship, the scholarship on the ethics of neuropsychiatric genomics, and the growing academic interest in young people. This thesis investigates the meaning that young people, i.e. genomic citizens in the making, attribute to the concept of biological citizenship, and its related notion of genetic responsibility, in the context of the application of genomics to psychiatric conditions. Because it falls under the tradition of empirical bioethics, this thesis draws normative conclusions about why and how the responsibilities that young people promote under their framework of biological citizenship should underpin and guide action in psychiatric genomics. Therefore, the primary questions of this research are as follows:

1. What meaning does the current generation of young people attribute to 'biological citizenship' in mental health settings?
2. What responsibilities do contemporary young genomic citizens promote under this account of biological citizenship?
3. Why and how should these responsibilities underpin and guide action about the use of genomic information in psychiatric settings?

To answer these primary research questions, it was necessary to engage young people in an empirical investigation. ASD was chosen as a case study because it has been the object of increasing interest of genomic research since twin studies in the 1970s; this has caused a range of ethical concerns, primarily due to the persistent disagreement on the way in which the condition should be

conceptualized – as a disorder, a disability, or a difference. Moreover, the implications of advances in autism genomics have so far been discussed with autistic people, their family members and autism professionals, while the publics, and young people in particular, have been completely excluded from this debate.

Thus, the empirical study investigated the moral attitudes that young people from the general population hold towards interventions that propose to use the knowledge of autism genomics to prevent or to treat the condition, or to intervene early to lessen autism risk. Because societal desire for these interventions is likely to be influenced by the way in which autism is conceptualized, a preliminary investigation of whether young people view autism as a disorder, a disability, or a difference was deemed necessary. This means that a range of secondary questions were developed to formulate the objectives of the empirical study, so that this could provide a picture of young people's views of autism and attitudes towards advances in autism genomics. Particular attention was paid to the arguments underpinning such views and attitudes to inform answers to the overarching questions of the present thesis. Figure 6 illustrates the link between the secondary questions and their contribution to answering the primary research questions.

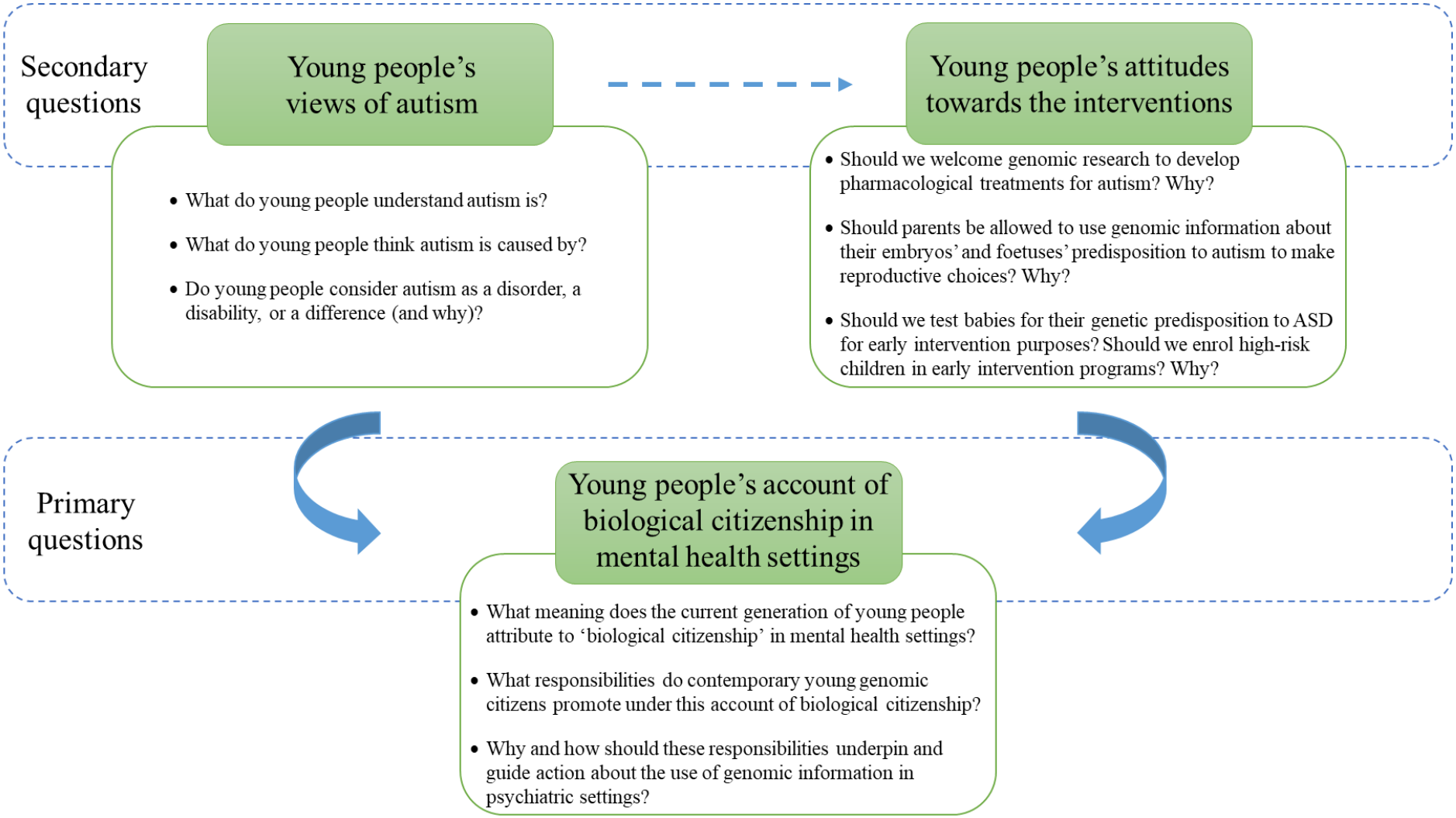


Figure 6: Primary and secondary research questions

This chapter provides a detailed account of the research methodology used to conduct this study. I will first discuss the epistemological foundations underpinning my chosen methodology (s4.2), including why I have conducted qualitative research; how my study falls under the tradition of empirical bioethics; and why I believe that a co-production model fits this research well. I will then describe the methodological design of this study (s4.3), particularly the consultation phase that I have conducted with the Neuroscience, Ethics and Society Oxford Young People's Advisory Group; the population and sample, and ethical requirements of my research; and the data collection and data analysis phases. I will conclude with some considerations around reflexivity (s4.4).

4.2. Theoretical issues

4.2.1. Rationale for qualitative research

Most handbooks on social science methods (see Bryman, 2016; Silverman, 2013; Sugarman and Sulmasy, 2010; Taylor et al., 2010) explain that, while quantitative methods are adopted when some previous understanding of a phenomenon exists and answer 'how many', 'how much' and 'how often' type of questions, the nature of qualitative approaches is discovery-oriented, descriptive and exploratory. Qualitative methods are used to explore a phenomenon of which there is little understanding, and they answer 'what', 'how' and 'why' type of questions, i.e. questions that could not be investigated by quantitative methods because they are not amenable to measurement (e.g. what experiences, understandings and attitudes certain individuals have, how they are formed and why). The goal of qualitative research is not to test hypotheses to generalize the study results (deductive method), but to better grasp the complexity of phenomena through an in-depth investigation that is hypotheses-generating (inductive method).

This brief introduction helps to clarify why I opted for a qualitative approach. In exploring young people's attitudes towards the use of genomic information to prevent autism at different life stages, I was not interested in identifying the proportion of young people who supported certain attitudes

towards autism genomics; rather, I aimed to achieve a deeper understanding of such attitudes by identifying the *underlying moral arguments*. This was part of a broader interest in discovering what moral concepts young people use at the intersection between genomics and psychiatry, of which there was little knowledge because no systematic research had yet investigated it. In this way, a side objective of my research was to collect empirical data to inform, albeit partially, theories about the way genomic citizens in the making reason around the present and other bioethics debates.

Thus, in my research I was fundamentally concerned about 'what', 'how' and 'why' type of questions that would ultimately inform an understanding of what young people's moral concerns tell about the meaning they attribute to biological citizenship in psychiatric settings, and the responsibilities they promote under their account. To this end, I adopted an active role, by challenging participants' contributions with counterarguments or asking them to compare autism with non-psychiatric conditions, in order to encourage them to better articulate their views. In section 4.3.4.2, I will justify why, among various qualitative data collection techniques, I opted for focus groups, to further clarify the epistemological commitments grounding my research design.

4.2.2. Empirical bioethics

This study falls under the tradition of empirical bioethics (EB), which emerged at the end of the last century, in part as a result of the social sciences critique of bioethics (Borry et al., 2005; Hedgecoe, 2004). According to this critique, bioethics was too abstract, dogmatic and distant from the particularities of the real world it investigated, i.e. healthcare contexts and their normative complexities, because it identified the authority of moral norms in rational justification and assumed that moral norms could be binding simply by virtue of their appeal to rationality. In this way, bioethicists addressed moral concerns through a top-down application of principles from moral theories to practice (Hoffmaster, 1994). Because morality is always socially embedded, i.e. morally loaded decisions are made within messy practical situations, bioethicists' theoretical analysis was accused of neglecting the human dimension of healthcare practice.

Against this critique, in finding solutions to normative questions *empirical* bioethicists' commitment is to engage with and be sensitive towards the peculiarities and nuances of the practical realities from which ethical issues arise (Strong, 2010), because only in this way can theoretical analysis (aspire to) make a difference in the real world (Ives et al., 2016). As it has been put, EB research that wishes to speak to real-world moral agents should 'address a normative issue that is oriented towards practice' (Ives et al., 2018). This is achieved by integrating empirical methods with ethical arguments, rather than conducting the empirical and normative analyses separately.

A problem, however, arises because since the 18th century, philosophers – the British empiricists, in particular David Hume – have contended that there is a radical distinction between 'is' and 'ought' claims or between facts and values. The philosophical literature on this issue is too vast to be appropriately covered here but, briefly, the argument is that ethical claims are distinct from factual claims because the former are normative (they prescribe how the world should be), while the latter are descriptive (they refer to how the world is). According to Hume, deriving an 'ought' claim from an 'is' claim is a logical fallacy (Hume, 1956, cited in McMillan, 2016). Another way of expressing the incompatibility between the empirical and the normative is to say that facts are objective and value-free, and values are revisable, subjective, and independent from facts; hence, values cannot be reduced to facts or natural properties, because there is something more in values that cannot be expressed in natural terms (G. E. Moore's naturalistic fallacy).

However, the 'is'/'ought' or 'fact'/'value' distinction does not necessarily mean that there cannot be empirical grounding for normative claims, not least because the epistemology of British empiricism has been largely criticized (McMillan, 2016). Some interpret the distinction as a warning that one needs to justify how to derive ethical claims from factual claims (Ives et al., 2016) – in this sense, EB adopts a non-empiricist and non-positivistic epistemology. The various methods that empirical bioethicists have theorized are different ways to integrate the empirical and the normative so that research can lead to empirically informed normative conclusions.

Broadly speaking, EB methods fall into two approaches (Davies et al., 2015). Dialogical approaches integrate normative analysis within the process of data collection. Normative conclusions are developed within the research encounter and their moral justification resides in the consensus-building process among research participants from which they derive. This approach gives priority to the stakeholders, while the researcher plays the role of a facilitator who communicates the knowledge (the normative claims) reached through a group process during data collection. Consultative approaches, instead, give priority to the 'thinker' (i.e. the researcher), who 'consults' participants to collect empirical data and then takes responsibility over the process of data analysis, which is conducted outside the research encounter, to develop normative claims. Rather than consensus, to justify the normativity of such claims this approach seeks theoretical soundness and some form of coherence (e.g. between empirical data and a moral theory).

However, these two umbrella approaches include methods with different epistemological and meta-ethical commitments, which produce different kinds of knowledge (Davies et al., 2015). Thus, empirical bioethicists need to clarify their position on the integration between the empirical and the normative, and so justify their adoption of a particular method (Ives et al., 2018). It is this task that I undertake below.

4.2.2.1. How does empirical bioethics apply to this research?

While developing my methodology I found my intuitions about ethics resonated with Dunn et al.'s idea that, to be oriented towards practice or to make a justified difference in the real world, empirical bioethics needs to develop 'arguments that are convincing, and that actually convince' (2012, p. 469). This means that an integration between moral–philosophical arguments and empirical arguments needs to be achieved because of the complementary nature of these kinds of arguments. The former aim to be binding by appealing to the rational and logical capacities of moral agents; however, logically convincing arguments may not convince agents because they may 'fail to resonate with one or more pertinent aspects of [their] lived experiences' (Dunn et al., 2012,

p. 470). The latter tend to be persuasive because they are based on a description of the concrete peculiarities of the context in which moral agents are, but they too need to be justified to guide action. Thus, under this perspective normative conclusions are justified only if they are the result of a normative analysis of empirical data that produces *coherence* between universal standards of justification for logically sound arguments and considerations of the nuances and particularities of the practical context (Davies et al., 2015).

In my study, the discussions with participants provided real-world data about young people's moral attitudes towards genomic advances in autism, and particularly the moral arguments underpinning their attitudes. This was informed by my theoretical framework (Chapters 1, 2, and 3): questions were framed based on the ethics literature on neuropsychiatric genomics, autism genomics, disability studies, etc. Therefore, data themselves touched upon ethical concepts (e.g. identity, neurodiversity, normalization, genetic determinism, stigma, etc.) on which a normative analysis could be done. Section 4.3.5 presents the analytic process through which I conducted such analysis. In the context of substantiating the epistemological commitments of my study, it will suffice to say that the specific research method that I used, which draws to some extent upon Dunn et al.'s grounded moral analysis methodology (2012), is a consultative and coherence-seeking method.

Being consultative, this method gives priority to the researcher, who owns the process of data analysis to reach the coherence described above. This process allowed me to reflect, outside of the research encounter, on how young people's contributions informed an account of the way in which they think of biological citizenship in psychiatric settings, since participants were not asked direct questions about these notions (see s4.3.4.2 below). In a consultative approach, the priority given to the researcher means that they play a central role in the co-construction of knowledge (Ives and Dunn, 2010). Like much social science and EB research (Dunn and Ives, 2009), the present study was indeed grounded on the ontological position of *constructionism* and the linked epistemological paradigms of *interpretivism* and *social constructivism*.

These theories include a variety of traditions, of which an accurate presentation is beyond the scope of this chapter; here I will only briefly explain how these paradigms informed my methodological choices. According to *ontological constructionism,* social realities are constantly co-constructed by social agents and their interactions (Guba and Lincoln, 2000). In my research, by exchanging arguments and counterarguments participants and I co-produced data about young people's views of ASD and attitudes towards advances in ASD genomics within the research process itself. Under this view and in contrast to the tradition of positivism, the social world cannot and should not be studied with the same methods as the natural world. On the contrary, social science research requires appropriate methods and tools to grasp the subjective dimension of social practices (Bryman, 2016).

In particular, it requires the subjectivist epistemology that characterizes *interpretivism* and *social constructivism* (Guba and Lincoln, 2000). Under these paradigms, empirical data are always partial representations of the social world, as they are dependent on sociocultural factors and on the contingent interactions between researchers and participants (Dunn and Ives, 2009), and multiple 'mental constructions' to apprehend the same social reality could be developed (Guba and Lincoln, 2000, p. 110). Therefore, knowledge of a certain social phenomenon (e.g. societal values around ASD and autistic people) should result from comparisons between different potential constructions, and interpretation and reinterpretation of empirical data. This allows to develop more accurate and sophisticated interpretations of the phenomenon, and bring categories and patterns identified in the data to a further level of abstraction (see s4.3.5).

Social constructivism raises methodological challenges, particularly around the credibility of a study result. In the social sciences researchers are faced with the question concerning 'how "data as construct", rather than "data as fact", can ever allow us to forge meaningful representations of the worlds we study' (Dunn and Ives, 2009, p. 93). Thus, the active role that the consultative method required me to play during data collection and analysis calls for some form of reflexivity on the

way in which my personal intuitions and experiences may have influenced the development of my normative conclusions. Some reflections around these challenges will be presented in s4.4.

It is important to note that, although drawing upon Dunn et al.'s account of empirical bioethics (2012), the methodology presented in this chapter diverges from it to the extent that this thesis does not aim to perform 'hard' normative work, whereby empirical data are used to develop a decisive conclusion or strong ethical argument that aims to convince the reader of adopting a specific position, e.g. on the 'right' way to conceptualize autism and the 'right' choices that biological citizens should make in the context of autism genomics. Rather, the aim of the philosophical and conceptual analysis of the empirical data performed in this research is to elucidate the moral concerns that young people raise about advances in autism genomics, in order to inform an understanding of the way in which they think of biological citizenship and genetic responsibility in mental health contexts.

To this purpose, the value of an empirical bioethics methodology that seeks coherence between universal standards of justification for logically sound arguments and considerations of the nuances and particularities of the practical context is that it allows to put participants' empirically derived accounts under philosophical criticism to identify where and why participants might promote arguments that fail to meet the universal standards of reasoning in moral argumentation. In turn, this allows researchers to assess whether participants' arguments should or should not be taken as sources to draw normative conclusions on how people should behave within a certain practice.

As I will show in the next chapters, in the present research such process allowed to distil, among young people's contribution, a 'better' way to address ethical issues about the use of genomic information in psychiatry, and so a 'better' understanding of the notion of biological citizenship in psychiatric settings, which could ground normative conclusions on the way in which young people's contributions should be used to guide choices at the intersection between genomics and mental health. Chapter 1 explained that in the present research putting young people's contributions

under normative scrutiny was essential not only to avoid that such contributions would be taken as a direct source for justifying certain practices in psychiatric genomics, notwithstanding how harmful they might be; but also to anticipate and address potential questions about why young people's insights, and the conclusions drawn by this thesis, should be taken seriously, given the persistent scepticism over young people's capacity to contribute to moral debates.

4.2.3. Co-production model

The development of the co-production model of research, also called participatory action research, responds to recent efforts of encouraging a 'stronger engagement between universities and a range of "publics"' (Banks et al., 2013, p. 265) to overcome the traditional separation and power imbalance between researchers and participants. Although I did not fully adopt this approach in all phases of my research, my study was informed by some of its principles.

According to scholars who support this model, a fundamental flaw of traditional research is that its design and undertaking require little (and often no) involvement from the groups on which the investigation is conducted (Beebeejaun et al., 2013). The aim of participatory research is to give ownership back to participants by seeking a collaboration between academic and non-academic stakeholders 'in the way research is conceptualized, practised and brought to bear in the life-world' (McTaggart, 1991, p. 171). In this way, research is conducted *with* rather than *on* participants (Beebeejaun et al., 2013; Heron and Reason, 2001; McTaggart, 1991), who cease to be the 'subject' of the research process to make a more active contribution.

Although its boundaries, advantages, and limitations are still contested (Seal, 2018; Wilkinson and Wilkinson, 2018), the co-production model aims at producing research with two core characteristics. First, the research process and design needs to be *participatory*, i.e. research participants should make a contribution to and have control over (all or some) phases of the research process (Durose et al., 2012). It is important to note that the term 'co-production' is used to refer

to research studies that seek stakeholder participation in different forms and to different extents, ranging from mere involvement (McTaggart, 1991), to contribution to all research phases, including setting the research agenda and methodology, and participating in data collection and analysis (Tandon, 1988). Second, participatory research is oriented towards *action* (Heron and Reason, 2001), i.e. it should aim to achieve 'transformational social goals' (Beebeejaun et al., 2013), or generate change within a certain social practice (Banks et al., 2013).

To this purpose, participatory research is based on an 'expanded' epistemology (Heron and Reason, 2008) that values different kinds of expertise, particularly researchers' academic expertise and participants' experiential expertise (Collins and Evans, 2002; Heron and Reason, 2008). These are considered as equally important and entitled to contribute to the production of knowledge (Beebeejaun et al., 2013; Durose et al., 2012). In fact, according to participatory researchers, research is enriched by integrating these kinds of expertise, because producing knowledge about and change in a certain social practice would be hard without appropriate engagement with the main stakeholders (Banks et al., 2013). As I will explain below, it is these claims around expertise and knowledge that motivated me to adopt some aspects of this model.

In the context of childhood research, the idea of co-production started to become popular after the publication of the United Nations Convention on the Rights of the Child (United Nations, 1989), which has led to the proliferation of studies exploring young people's perspectives and involving children as 'co-researchers' (s2.4.1). However, the idea that studies seeking young people's increased participation produce necessarily 'better' research is contested (Durose et al., 2012; Holland et al., 2010; Nolas, 2011; Seal, 2018). Two concerns are particularly worthy of note.[20]

[20] Other concerns regard the risk of potential tokenism; the lack of systematic evidence on whether children themselves desire and seek participation; challenges arising when young people's preferences clash with those of the researchers; and others (Birch and Miller, 2002; Holland et al., 2010; McLaughlin, 2006; Pavarini et al., 2019).

Holland et al. (2010) suggest that participatory research could reinforce those patronizing assumptions around children's vulnerability and incompetence that it seeks to contest, because it might encourage the idea that it is the adult researcher who empowers children or enables their agency, by making them co-investigators in research. I believe that my argument about young people's capacities to contribute to moral debates (s2.4), and my reflections on how working with young people has influenced and improved my methodology, of which this chapter is rich, show my genuine interest in young people's contribution and effort to reduce any occasion of power imbalance that could have arisen in my study.

A second criticism is that participatory research is not as innovative a methodology as it claims to be, because many research stages are still very much conceptualized, designed and implemented by researchers, who often use well-established methods to engage children (Gallacher and Gallagher, 2008). On this point, I believe that it is the researcher's responsibility to clarify how and to what extent young people's participation is sought in their studies and to make this available for scrutiny by other researchers. This is the aim of the remainder of this section.

My study was part of an umbrella project called *Citizens: Early Intervention Ethics*, which is led by Professor Ilina Singh.[21] This project is committed to involving young people in bioethics debates to understand their values about and moral attitudes towards psychiatry and innovations. In sharing the commitment of the *Citizens* project, I was convinced of the value of integrating two forms of expertise within my study: (1) my academic and philosophical interest in young people's capacities to contribute to moral debates (Chapter 2); (2) young people's direct experiences with engaging in moral discussions and their experiential knowledge of the tools that encourage children's engagement or probe their contributions.

[21] *Citizens* is supported by Professor Singh's Wellcome Trust Senior Investigator Award for a project entitled *Becoming Good: Early Intervention and Moral Development in Child Psychiatry* [104825/Z/14/Z]. Although part of *Citizens,* the present study on autism was supported by a Wellcome Trust Doctoral Studentship that I was awarded independently for a project called *An Empirical Study of Children and Adolescents' Perspectives on Testing Minors for their Genetic Predisposition to Psychiatric Disorders* [203329/Z/16/Z].

I considered the integration of these two kinds of knowledge as particularly important because of the complexity of the scientific and ethics topics discussed in my study, which meant that my research methods had to be informed by young people's contribution to ensure that my research would speak to and would be understandable to my target population. In other words, I was convinced that young people could engage with complex scientific and ethical concepts, and that as a researcher I had the responsibility to develop the appropriate methods and tools to elicit young people's views, so that their voices could be heard.

These background considerations found in the conceptualization of participatory research an appropriate model for the development of the present study, particularly for consulting young people on the most appropriate methods to use. However, as I will show in the discussion of the consultation that I conducted with the Neuroscience, Ethics and Society Oxford Young People's Advisory Group (s4.3.1), a participatory approach that was originally thought to inform my methodological choices turned out to make a substantial contribution to conceptual aspects of my study. Of note, I did not seek young people's participation in all research phases, which is why my study was *informed* by the co-production model, rather than fully adopting it. Differently from other examples of participatory research (see Nolas, 2011; Warming, 2006), children who were involved in the consultation phase of my research did not take part in my study as participants.

Having discussed the conceptual commitments of my methodology, in the next section I will provide further details on how the chosen research design and methods fit the issues laid out in my background chapters, and so it is appropriate to answer my research questions.

4.3. Methods

4.3.1. Consultation phase: The NeurOx Young People's Advisory Group

The Neuroscience, Ethics and Society Oxford Young People's Advisory Group (NeurOx YPAG) is a group of (originally) 47 14–18-year-olds from various backgrounds, with or without mental

health diagnoses, which I contributed to set up and I co-led until January 2019. In February 2017, members of the NeurOx YPAG were selected via an application process from state and private schools across Oxfordshire for their interest in ethics and mental health. The group was established to support the research conducted by the Neuroscience, Ethics and Society (NEUROSEC) team, particularly the *Citizens* project, and it meets every term to advise and collaborate on all stages of our team's studies. Although advisory groups that include patients, research participants and the public have become increasingly common to conduct participatory health research with youth, to our knowledge this is the first YPAG in Oxfordshire, and the first one ever focusing on ethics (for a detailed account of the NeurOx YPAG model, see Pavarini et al., 2019).

Because YPAG members had a range of backgrounds and lay expertise on mental health, and because we made efforts to include in the group marginalised young people, those with part-time jobs and individuals with various skills (see Pavarini et al., 2019), I considered the NeurOx YPAG diverse enough to be representative of those children from the general population that I would have encountered in my focus groups. Thus, I concluded that a consultation with the YPAG would be helpful in framing and designing a study involving such population.

In my research, YPAG members first took part in a range of brainstorming exercises on autism and its causes, which allowed me to assess young people's familiarity with autism and their capacities to reflect on ethical issues around ASD. This was important not only from a pragmatic point of view – I expected that increased familiarity with a certain psychiatric condition would make young people more interested in enrolling in a study about it and more confident in their capacity to contribute to the discussion; but also because it contributed to the selection of autism as a case study in my research. Although autism is among other psychiatric conditions whose biological and genetic underpinnings are being investigated to improve risk calculation and prevention, my hypothesis was that young people may be more familiar with it than other psychiatric conditions. Indeed, since the late 1990s autism has acquired a high profile in the public debate (Fitzgerald,

2012), in part thanks to autistic people's and their families' activism around the value of autistic traits (Cascio, 2012). More recently, the climate activist Greta Thunberg has publicly spoken about her Asperger's diagnosis (Thunberg, 2019), and public discourse around autism has been seen in media representations targeting a young audience, e.g. the Netflix series 'Atypical' (Rashid, 2017).

In addition, in England more than 70% of children on the spectrum attend mainstream schools (Great Britain. Department for Education, 2017); and ASD is the primary type of need for pupils with an Education, Health and Care plan, who therefore require more than Special Educational Needs (SEN) support (Great Britain. Department for Education, 2020). Thus, it is likely that young people will have heard of autism because of classmates who need extra support in various classroom situations. Despite the widespread discourse around ASD, so far children's voices have been absent in the public debate, which makes this research relevant to contribute to move the debate forward.

All YPAG members who took part in the sessions on autism had knowledge of ASD and most knew someone on the spectrum, while from other activities conducted with the YPAG I had evidence that many had little familiarity with other psychiatric conditions such as psychosis, on which I had initially focused my research. The brainstorming activities also showed that young people could reflect around ethical issues in the context of autism and autism genomics, such as the debate about the nature of autism, or whether autism should be treated or 'cured'. However, it became clear that YPAG members were less familiar with risk factors for autism, which meant that I had to carefully plan the part of my focus groups on ASD aetiology (see s4.3.4.2).

The main contribution of the NeurOx YPAG was in shaping the appropriate methods to discuss the complex scientific and ethical concepts of this research with a young population of participants. This was possible thanks to the participation of two groups of five YPAG members, one of which had not taken part in the initial activities on autism, in a pilot of my focus group discussions (FGDs). In qualitative research, pilot testing is important to reveal problems within the topic guide (e.g.

whether participants may misunderstand some questions); assess whether some activities need to be reconsidered; determine whether participants' interest and attention may decrease at certain junctures; or test how long administering a topic guide takes (Bryman, 2016; Taylor et al., 2010).

In my work with the NeurOx YPAG I experienced these and other advantages of conducting pilot testing. Of particular interest is that the pilot revealed that some activities needed to be rethought. It became clear that the original topic guide required too much reading of scenarios, which made participants tired and increased the risk of losing their attention and interest. Many of the interactive tasks that I used during data collection (see s4.3.4.2) were developed thanks to the outcome of the pilot study and to the YPAG's input during subsequent sessions. For instance, the YPAG helped me select some YouTube video clips on autism to use as educational material with my research participants. Moreover, after the pilot I added space in my topic guide to recap what is currently known about autistic traits and risk factors for autism, because some YPAG members' contributions highlighted the importance of ensuring that participants would not leave the discussion with significant misconceptions about autism.

Although I had conceived the pilot testing more as a way to inform my methodological design than to test what themes emerged from the discussion, some of the YPAG members' ideas made substantial intellectual contributions to the theoretical framework of my study. Before conducting the pilot, I had categorized ethical concerns around autism genomics as issues in either research or clinical settings, because of influence from the ethics literature on genomics (in general, and in the context of autism in particular, see Hens et al., 2016a). However, the pilot discussion highlighted that, because of the centrality of the concept of identity in ethics discussions about autism, the normative relevance of autism genomics may not lie in the distinction between research and clinics, but in interventions performed at different life stages. Some YPAG members argued that interventions after birth, especially later in life, are more ethically problematic because they affect individuals whose identity has already been constructed around the ASD diagnosis. Thus, although

the ethical issues in autism genomics discussed in my study remained essentially the same, I re-conceptualized them around a framework (presented in Chapter 3) that focuses on prevention before birth, interventions in early years and treatments later on in life. This shift was essential to rephrase the secondary research questions of my study to their current version.

Finally, the YPAG reviewed recruitment materials, including information sheets, consent and assent forms, and invitation letters, and made recommendations of charities or youth groups where to recruit participants. The overall experience with the NeurOx YPAG contributed to build my confidence in working with children and helped me become accustomed to discussing complicated topics at the intersection between genomics and psychiatry with a lay audience. In this way, I learned to develop questions in a language that was age-appropriate and could elicit relevant responses, which contributed to ensure the reliability of the study findings (Singh and Keenan, 2010).

4.3.2. Population and sample

4.3.2.1. Population

To be able to take part in my research participants had to be between 13 and 18 years old and have good level of spoken English. The lower limit of my population's age range (13 years) was chosen because, according to the English National Curriculum, by the end of Key Stage 3 (Year 9, when pupils are 13–14 years old) students become familiar with the basic concepts of genetics, e.g. genes, chromosomes, DNA, inheritance, difference between species and biodiversity; by the end of Key Stage 4 (Year 11) they learn about more complex concepts, such as gene–environment interaction, multifactorial traits, and dominant and recessive phenotypes, and they discuss some ethical issues arising from advances in biotechnologies (Great Britain. Department for Education, 2013).

I did not set any criteria to exclude young people with autism or with autistic relatives, which allowed me to recruit a population with a range of lay expertise on autism. However, I did not

specifically sample for these two groups. Indeed, for reasons explained in Chapter 1 (s1.1 and s1.2.3) my study investigated the views and attitudes of the general population, and thus it did not have a specific focus on young people's personal or family experiences of autism. This means that from a research ethics perspective there was no strong rationale for only involving autistic young people or their siblings, who might be considered as a more vulnerable population.

Moreover, time and resource constraints had to be taken into account. A study set out specifically to recruit a clinical population of autistic young people would have required me to obtain NHS ethics approval, which is notably a time-consuming process that would have had to be added to the redesign of the focus of my research at the beginning of my second year. In addition, there are no approved methods to conduct qualitative research with autistic people, particularly those with lower IQ and significant communication difficulties (Elsabbagh et al., 2018), and so my population would have been biased towards high-functioning people. Finally, a study that asked siblings or close relatives of autistic individuals to comment on their family experience and reason around ethical implications of ASD genomic advances may have been quite emotionally demanding for my participants. Since I had no experience of empirical research prior to my doctorate, after contemplating this option with my supervisors we concluded that, without appropriate training, I would have found managing potentially sensitive situations with these participants too challenging.

4.3.2.2. Recruitment and study sample

Participants were recruited via schools, Young People's Advisory Groups, and other youth groups in Oxfordshire, London and Birmingham. In order not to interfere with school activities, recruitment in schools occurred from Year 9 and Year 12, i.e. years in which pupils are not busy with the General Certificate of Secondary Education (GCSE, Year 11) and A-level exams (Year 13).

I followed a similar recruitment procedure in the various groups. I first gained permission of the site for my study by approaching a site gatekeeper (e.g. school teacher or youth group leader) via

email or phone. Once permission was gained, the gatekeeper sent the parents or legal guardians of young people under 18, or 18-year-old participants themselves, an invitation email to take part in the study (Appendix B), including a link to an online Parent/Legal Guardian Information Sheet and a link to an online Young Person Information Sheet (Appendix C), which explained the purpose, objectives and design of my study[22]. Participating sites were asked to advertise this study on their websites, newsletters and social media feeds. Moreover, study posters were placed within the Department of Psychiatry at the University of Oxford and online advertisements were posted on my personal or the NEUROSEC team's social media platforms (website, Twitter and Instagram). The study ads included a link to the Oxford Department of Psychiatry's 'Get involved' webpage, where potential participants could read more about the study and find my email address to request further information. Examples of advertising materials are shown in Appendix D. On one occasion, I presented my study at the participating youth group's meeting to draw potential participants' interest.

Both sites and people were recruited via non-probability forms of sampling. Sites were selected via purposive sampling, since the goal was to approach settings where young people within my study age range could be easily found (Bryman, 2016). I first approached schools to ask for their interest in being involved in my study; however, this recruitment strategy proceeded slowly. Out of nine schools contacted between March and June 2018, four schools declined the opportunity, three schools never replied and only two schools accepted to be involved. The main reason for declining was that, despite their interest in the research topic, teachers were too busy to commit to the organization of the focus groups in their schools, particularly during the summer term (April–July

[22] Please note that the invitation emails and information sheets specified that: 'Researchers think that a combination of genes and environmental factors may cause autism'. This was meant to contextualize the focus of the present study on young people's attitudes towards using genomic advances to predict, prevent, and treat autism. After considering whether this sentence could limit the validity of the findings of the present study, which included questions about young people's understanding of risk factors for autism, I concluded that it was unlikely that such piece of information would influence potential participants' pre-existing knowledge and assumptions about the causes of autism.

2018). This is why youth groups were initially prioritized over schools to identify research participants.

Recruitment of participants started by convenience sampling, which is an acceptable technique in an exploratory investigation (Pearlman and Starks, 2010). Thus, the first FGDs consisted of young people who were already undertaking other activities within the NEUROSEC team, and young people approached thanks to formal and informal contacts that my team had with leaders of other UK YPAGs or youth groups (Pavarini et al., 2019). Recruitment via youth groups proved to be more successful in the summer months, as I was able to recruit at least a few participants in four out of five youth groups contacted between May and August 2018. In the unsuccessful case, failing to recruit participants was primarily due to time constraints, which made it difficult to obtain parental consent ahead of the scheduled meeting. Moreover, a snowball sampling technique was used, whereby members of the NeurOx YPAG and work experience students in our team were asked to personally tell friends about the study, by pointing them to information posted on the NEUROSEC team's social media platforms.

Fifty-six participants were recruited in total. Table 3 shows a breakdown of study sites where participants were recruited, as well as participants' distribution by age range in each site.

Table 3: Sample sizes by study site and age range

Study site	**13-15 years old N**	**16-18 years old N**	**Total N**	**%**
YPAG 1	3	4	7	12.5
YPAG 2	1	3	4	7.1
YPAG 3	0	4	4	7.1
Youth Forum 4	1	1	2	3.6
School A	5	0	5	8.9
Foundation School B	5	5	10	17.9
Academy C	7	2	9	16.1
Science Partnership D	0	4	4	7.1

School E	6	0	6	10.7
Others*	0	5	5	8.9
Total N	28	28	56	
%	50	50		
*These include young people who were already undertaking activities within our team and young people who contacted the research team after seeing online advertising				

4.3.3. Ethical considerations: Research involving minors

This study was reviewed by the University of Oxford Central University Research Ethics Committee (CUREC; Ref number: R56389). I obtained ethics approval in March 2018, but I had to subsequently seek amendments to be able to conduct the study in schools outside of school hours; to extend recruitment outside schools and conduct FGDs within the University of Oxford or Oxford Colleges, or at charities or youth groups' meeting venues; and to use new recruitment documents that could be distributed to various locations such as sports centres, youth group venues and social media platforms. The third and last amendment was granted in July 2018, when I started data collection. Appendix E includes the CUREC original approval letter and approval of amendments. Some ethical requirements that I had to comply with (e.g. confidentiality, personal data protection, and study data storage and destruction) are common to any research study, while the main ethical issues that this study specifically raised concerned the combination between the involvement of minors and the sensitivity of the topic discussed.

Minors have traditionally been considered as a 'vulnerable' group in research, and so ethics committees tend to require researchers to meet higher ethical standards when involving children. Leaving aside the criticisms raised towards the assumed vulnerability of children (s2.4), this means that studies involving minors tend to be time-consuming (Singh and Keenan, 2010). Even though the study procedures involved minimal risk, CUREC considered my research topic of a sensitive

nature, and thus 16–17-year-olds could not consent for themselves;[23] rather, parental consent had to be obtained for all participants under the age of 18. Parents or legal guardians were asked to provide consent for their children's participation by completing an online consent form integrated in their information sheets (Appendix C). Young people were asked to fill out an online expression of interest form, which was integrated in their information sheets (Appendix C). Assent from participants under 18 and consent from 18-year-olds were then taken in person before the FGD began (Appendix F).

The process of obtaining consent from parents/legal guardians, getting young people to complete their online expression of interest forms and taking care of the logistical organization of the FGDs did not proceed smoothly, meaning that many follow-up emails had to be sent in most cases. Thus, collaboration with the study site gatekeepers and gatekeepers' enthusiasm about my research were essential for the successful organization of the FGDs. This was particularly evident in the second phase of recruitment in schools, which was more successful because I could rely on contacts that my team had with staff in various schools. When this was missing, the process often resulted in last-minute cancellations of planned FGDs due to lack of an adequate number of participants.

In addition, during the planning phase of my study I decided not to conduct this research with young people on the spectrum or their siblings for reasons outlined in s4.3.2.1. In the absence of a direct and personal investment in the research topic, the general population of young people may have been less interested in signing up to the study. Moreover, CUREC only allowed a £5 book token as a reimbursement, which I had to email to parents or legal guardians if participants were under 18. This was considered the most I could provide in order not to offer an undue incentive, and it could only be provided to participants recruited outside schools, so not to introduce differences between schoolmates. Because the study required significant time commitment (about two hours of group discussion), some young people, particularly those without a specific interest on ethics, autism, or

[23]https://researchsupport.admin.ox.ac.uk/files/bpg04competentyouthspdf-0

genomics, or those who could not afford the time commitment for a small reimbursement, may have gauged the study not worth participating in.

It is difficult to quantify how this might have impacted recruitment. Some young people may have nonetheless been motivated to participate by the prospect of learning about autism – and indeed I often received emails from participants or their parents who expressed how much they had enjoyed the opportunity and had learnt from participating. However, these considerations seem to confirm what has already been argued by others, i.e. that 'the problem of access is, unfortunately, a major barrier to research with vulnerable or marginal populations, and it requires planning and effort to overcome' (Singh and Keenan, 2010, p. 702). This made the empirical phase of my research more time-consuming than I had planned. Nonetheless, I believe that time commitment, which is one of the main reasons why in clinical and social research we tend to lack data collected from young participants, should not discourage researchers from involving minors in research. Rather, important methodological lessons can be learned to inform future research studies with young people.

In this research, building trust and a supportive relationship with the study site gatekeepers was important to obtain permission to conduct the study, and subsequently to make sure that parents or young people who had expressed their interest in participating would be responsive to emails concerning the logistics of the FGDs. Strong working relationships were sometimes initiated by simply having a phone call or an in-person meeting, rather than emailing the gatekeeper about the study; however, as aforementioned more often I found it easier to gain the gatekeeper's trust if I had gotten in touch with their site through personal relationships (e.g. colleagues or friends working in similar fields).

Moreover, because it seemed that the stricter ethical requirements about research involving young people made it difficult to access participants, and increased the chances for last-minute cancellations, throughout the course of the empirical phase I learned that I needed to have more

groups lined up, in order not to miss opportunities for data collection. Finally, some have argued that participatory research methods could be useful to overcome challenges about involving minors in research (Morrow and Richards, 1996). In my study, having the opportunity to consult the NeurOx YPAG was extremely helpful to identify the most appropriate platforms where to advertise my study to reach young people. Moreover, the NeurOx YPAG's input was essential for me to learn how UK schools organize their terms, and so identify times during the year in which school staff would be less busy and more willing to support my study. Other challenges about involving young people, which are not strictly related to ethical requirements, are discussed in s4.4.

4.3.4. Data collection

Data collection was conducted in two phases. Phase I lasted from the beginning of July 2018 to the end of August 2018. It was followed by a preliminary analysis of the data collected up to that point in order to inform Phase II, which lasted from October 2018 to March 2019. This study involved the collection of two sets of data, i.e. participants' demographic questionnaires (s4.3.4.1) and focus group discussions (s4.3.4.2).

4.3.4.1. Demographic questionnaires

At the beginning of each FGD, participants were asked to complete a questionnaire, which included questions about their demographic characteristics; their favourite subject in school; and their direct or indirect experience with autism, i.e. whether they had been diagnosed with ASD or knew anyone on the spectrum, and how often they saw them (Appendix G). These questions were meant to assess how varied my sample was, including considerations about whether the study had drawn the interest of young people who are passionate about a certain school subject (e.g. science; personal, social, health and economic education) and the sample's range of expertise on autism. Participants' demographic characteristics are shown in Table 4.

Table 4: Participants' demographic characteristics	
Characteristic	**Overall sample N, (%)**
Age	
13-15 years old	28 (50)
16-18 years old	28 (50)
FGD (Age)	
FGD, 13-15 years old only	4 (33.3)
FGD, 16-18 years old only	5 (41.7)
FGD, Mixed age	3 (25)
Gender	
Female	39 (69.6)
Male	17 (30.4)
Other	0 (0)
FGD (Gender)	
FGD, Female only	4 (33.3)
FGD, Male only	1 (8.3)
FGD, Mixed gender	7 (58.3)
Ethnicity	
White British	34 (60.7)
Other White Background	4 (7.1)
Mixed White and Asian	4 (7.1)
Others	14 (25)

Fifty per cent of participants were from the younger age range (13–15 years old). Only three FGDs included participants from both age ranges, while five groups had only older participants and four only younger. Thirty-nine females and 17 males participated in this study, and most groups had members from both genders. Most participants were White British (61%), and the next two most represented ethnicities were Other White Background (7%) and Mixed White and Asian (7%).

Table 5 shows participants' favourite school subjects and familiarity with autism.

Table 5: Participants' school interests and familiarity with autism	
Characteristic	**Overall sample (N= 56), n (%)**
What is your favourite subject in school?*	
Science	23 (41.1)
Art and design	10 (17.9)
English	7 (12.5)
Mathematics	4 (7.1)

Other	18 (32.1)
Have you ever heard of autism or Asperger syndrome?	
Yes	54 (96.4)
No	2 (3.6)
Prefer not to say	0 (0)
Have you ever been clinically diagnosed with autism or Asperger syndrome?	
Yes	0 (0)
No	55 (98.2)
Prefer not to say	1 (1.8)
Do you know anyone who has autism or Asperger syndrome?	
Yes	47 (83.9)
No	9 (16.1)
Prefer not to say	0 (0)
Please specify your relationship with them*	
Friends	28 (50)
Cousins	7 (12.5)
Siblings	5 (8.9)
Schoolmates	5 (8.9)
Other	3 (5.4)
Not applicable**	13 (23.2)
How often do you usually see the people you know with autism or Asperger syndrome?*	
Once or twice a year	7 (12.5)
Once or twice a month	14 (25)
Once or twice a week	13 (23.2)
Every day	14 (25)
Other***	1 (1.8)
Not applicable**	8 (14.3)
*Respondents were permitted to give more than one answer. As a result, numbers exceed the total sample size **Either did not know anyone or did not answer this question ***One participant did not select any given option but wrote 'once every few years'	

Science was a shared interest among many participants, who might have been attracted by this study because it related to the topic of genetics. All participants said that they had never been diagnosed with ASD, except for one young person who ticked the option 'Prefer not to say'. However, most participants (84%) personally knew someone with autism, including friends, cousins, siblings (mostly brothers) and schoolmates, and they tended to see them fairly often, with

'once or twice a month' (25%), 'once or twice a week' (23.2%) and 'every day' (25%) as the most chosen options. Although this does not signal that they necessarily had a close relationship with the people on the spectrum they knew, it suggests that participants were to some extent familiar with the topic of autism.

These characteristics did not appear to systematically account for differences in participants' responses. Data analysis identified only a few examples where age and familiarity with ASD seemed to influence participants' knowledge of and arguments about genomics and autism (see Chapters 5 and 6). Although most participants could engage in complex reasoning about ASD aetiology and felt confident about articulating explanations for their intuitions, the younger participants tended to use less accurate language, which at times revealed deterministic views. Moreover, on some occasions participants deferred to those group members who seemed to know more about autism, usually because of their close familiarity with people on the spectrum.

4.3.4.2. Focus groups

Empirical data on young people's views of autism and moral attitudes towards advances in autism genomics, and the arguments underpinning such attitudes, were collected through focus groups, which were audio recorded and transcribed. I conducted 12 FGDs, four in Phase I and eight in Phase II. Most focus groups included between four and seven participants[24], which falls within what some scholars have described as the 'ideal' focus group size (i.e. between four and eight people, see Kitzinger, 1995, p. 301). As a result of the challenges I encountered in accessing and recruiting participants (s4.3.2.2 and s4.3.3), two FGDs involved two and three participants respectively. FGDs lasted between one and two hours. They were conducted in a quiet and private room within schools, the University of Oxford, Oxford Colleges, libraries, or youth groups' meeting venues.

[24] Breakdown was as follows: four FGDs with four participants, three FGDs with five participants, one FGD with six participants, and two FGDs with seven participants.

Justification for conducting focus groups

Several considerations supported the use of focus groups as the most appropriate data collection technique for the aim of my study. First, FGDs simulate 'naturally occurring' conversations among research participants (Kitzinger, 1995, p. 300), especially within pre-existing groups, as most groups in my study were. In focus groups (at least as understood within this research) participants are seen as 'social beings co-constructing meaning' around a social phenomenon (Belzile and Öberg, 2012, p. 461). In this way, FGDs provide the context for the everyday discussions that 'shape our attitudes, routines, and behaviours' (Barbour, 2010, p. 337). This context is well suited to investigate how society values autism and autistic people, and how the publics think these values should guide the use of ASD genomic information for the primary and secondary prevention of autism.

Moreover, FGDs may help participants feel at ease because, in contrast to one-to-one interviews, no one individual is put under pressure to answer researchers' questions (Barbour, 2010). I considered this point very important in the context of the present study, as there is evidence that young people consider genetics a difficult topic to discuss (Iredale et al., 2010). Furthermore, in a group with their peers young participants may be less intimidated by the researcher and so the discussion may be more youth-led. Barbour (2010) also suggests that, because any one participant is not constantly accountable for answering questions, FGDs may be a particularly good fit for studies exploring topics that are not directly relevant to participants, as might have been the case for participants from the general population. The group setting might have provided them with enough time to reflect on ethical issues to which they might never have paid particular attention.

Finally and most importantly, in FGDs participants' interaction is part of the study method (Clark, 2009; Kitzinger, 1995). Because the group dynamic stimulates discussion of potentially opposing ideas, participants are compelled to articulate a justification for their arguments, question their own assumptions by listening to others' contributions and possibly change their perspectives (Barbour,

2010; Kitzinger, 1995). Thus, FGD participants form their own knowledge in dialogical relation with the other participants and with the researcher (Belzile and Öberg, 2012). Moreover, when differences of opinions emerge in focus groups, the researcher has the opportunity to understand why they exist *in situ* and together with participants; data collected by interviews might instead require researchers to do 'armchair theorizing' to ascertain the reasons underpinning disagreements among interviewees (Kitzinger, 1995, p. 301). In this way, focus groups are best placed 'to examine not only what people think but how they think and why they think that way' (Kitzinger, 1995, p. 299), which was my research aim (s4.2.1).

Indeed, in my facilitator role I specifically encouraged interaction by allowing group members to exchange stories, experiences and questions (Duggleby, 2005; Rothwell, 2010), and I played an active role to investigate what young people's moral attitudes towards advances in ASD genomics were and why. By proposing counterarguments, I facilitated moral reasoning and argumentation among my participants and encouraged them to articulate the arguments underpinning their moral attitudes. In this way, I took advantage of the group dynamic for the purpose of my research, which would not have been possible in individual interviews. Such an active role also allowed me to ensure that voices of dissent were not silenced but welcomed (Kitzinger, 1995), and to prevent the replication of pre-existing social dynamics and relationships between children in the research encounter, hence minimising the chances that some voices would dominate the debate (Smithson, 2000).

The rationale and content of the FGD topic guides

The focus groups in this research were divided into two parts: Part 1 focused on participants' views of autism, while Part 2 focused on their attitudes towards advances in autism genomics. Thus, each FGD involved the discussion of two different topic guides, which were quite structured because I was aware of both the complexity of the research topic and the widespread scepticism about young people's capacity to contribute to moral debates (Singh, 2016a). The topic guide for each part of

the FGDs was divided into three activities, which explored different nuances of young people's understanding of autism or moral attitudes towards autism genomics. To make sure that the empirical data could ultimately help me answer the primary research questions, each activity was focused around one of the secondary questions listed in Figure 6 at the outset of this chapter.

The activities were then divided into tasks, which either probed the same topic in different ways (such as young people's understanding of 'neurodiversity') or investigated different ethical concepts (e.g. genetic essentialism, genetic determinism, stigma, etc.). Because the pilot study with the NeurOx YPAG had highlighted the importance of interaction within the group (s4.3.1), the various tasks had an interactive nature. My topic guides slightly evolved over time, and Appendix H shows the final versions.

Part 1 served a quasi-educational function. Given the complexity of the research topic, and the involvement of participants who did not necessarily have any direct relation to autism, I deemed it important to ensure that young people could obtain a certain level of understanding of and familiarity with the complexity of autism in Part 1 to feel comfortable about discussing the ethics of interventions to prevent autism at different life stages in Part 2. These preliminary exercises also created the opportunities for me to address any misconception about autism that participants might have had. Table 6 summarizes the activities undertaken and topics covered in Part 1, including the specific secondary questions that they aimed to address.

Table 6: Activities and tasks in Part 1: Young people's views of autism

Activity	**Tasks**
Understanding autism: What do young people understand autism is?	• *Brainstorming activity on autism:* Can I start off by asking what you know about autism? Do you personally know anyone who has autism? • Please comment on this animated video on autism: https://www.youtube.com/watch?v=7JdCY-cdgkI • *Brainstorming activity on ASD spectrum:* 'If you have met one person with autism, you have met one person with autism'. What do you think this quote means?

Understanding the aetiology of autism: What do young people think autism is caused by?	• *Brainstorming activity on ASD risk factors:* What do you think causes autism? • *Participants were given a magnet, and shown a whiteboard with the letters E (environment) and G (genetics) written on two opposite sides of the whiteboard; a line linked the two letters.* As a group, please place this magnet on the line to say whether you think that autism is caused primarily by genetics or by the environment. • *Participants were given one set of magnets displaying the following items: 'genes', 'having older parents', 'vaccines', 'exposure to high level of testosterone in the womb', 'complications during pregnancy or birth', 'diet', 'cold mothers', 'bad parenting', 'having older siblings with autism'.* On these magnets you can read factors that may or may not increase people's risk for ASD. As a group, please place them on the whiteboard under 'Yes', 'No', or 'Question mark', depending on whether you think that they may contribute to ASD or not.
Understanding the nature of autism: Do young people consider autism as a disorder, a disability, or a difference (and why)?	• *Participants were shown a graph to visualize that autistic people tend not to have a typical average level of abilities across different tasks, but capacities that are at the extremes.* Would you say that someone who has all these traits has a disorder, a disability, or just a different personality? Why? • *Brainstorming activity on neurodiversity:* Have you ever heard of 'neurodiversity'? What are the implications of this concept on the way we should treat people with autism?

The first activity was a brainstorming exercise about autism and autistic traits, in which I investigated young people's *understanding of autism*. If participants seemed not to know much about autism or if the group was quiet, they were shown an animated video clip on ASD to comment on. Moreover, if they did not do so spontaneously, they were asked to reflect on the spectrum nature of autism. During data collection I realized that the video clip in Table 6, which provides a very positive idea of autism, could have hindered participants from mentioning any negative traits that they associated with autism. After the second focus group, I therefore decided to show different videos depending on whether the group had mostly discussed ASD challenges, in which case I

selected the video clip in Table 6, or positive traits, in which case they were shown a clip on ASD difficulties;[25] no video clip was used if participants were knowledgeable about autism.

The second activity focused on young people's beliefs about the *aetiology of autism*, which I had anticipated could inform participants' discussion about the genetic-based interventions for the primary and secondary prevention of autism in Part 2. In addition to a brainstorming exercise about causes of autism, two tasks were undertaken. Participants were asked to express whether they thought that autism is mainly caused by genetics or by the environment, using a visual analogue scale (gene–environment; see Table 6). They were then involved in a sorting-out exercise in which they had to decide whether some environmental factors could cause autism (see Table 6).

These introductory activities about ASD and ASD aetiology were meant to help participants engage with the research topic so that they could discuss how they conceptualized the *nature of autism*. This was assessed in the third activity, when participants were reminded of the strengths and difficulties that they had associated with autism, and then asked to consider whether, based on those traits, they saw autism as a disorder, a disability, or a difference, thereby positioning themselves in the debate about the different models of autism (Chapter 3). This discussion allowed me to introduce and investigate young people's familiarity with the concept of neurodiversity. A minority of groups (two out of 12) that were unfamiliar with this concept were asked to read and comment on a leaflet on neurodiversity produced by colleagues at the University of Exeter (Appendix I). Young people were also interrogated on their views on the neurodiversity movement's argument about accommodating and celebrating ASD traits, which in turn served as a way to move to the ethics discussion about interventions to prevent ASD at different life stages. Thus, the discussion about neurodiversity allowed the shift from activities focused on young people's factual knowledge of ASD to those that increasingly investigated their normative take on the research topic.

[25] These were either http://www.healthtalk.org/peoples-experiences/autism/life-autism-spectrum/making-friends-social-life-autism on social-communication difficulties, or https://www.ambitiousaboutautism.org.uk/hidden/my-sensory-world-by-louis-morel on hypersensitivity.

Part 2 of the FGDs discussed whether and how genomic information should be used for the prediction, prevention and treatment of autism, and focused on the three sets of genomic-based interventions introduced in Chapter 3: (1) pharmacological treatments targeting specific risk markers or subcategories of people with the same genotype; (2) prenatal diagnosis (PND) and preimplantation genetic diagnosis (PGD) to prevent autism before birth; and (3) genetic tests performed on young children to assess their susceptibility to autism for early intervention purposes. Table 7 summarizes the activities undertaken and topics covered in Part 2, including the specific secondary questions that they aimed to address.

Table 7: Activities and tasks in Part 2: Young people's attitudes towards the interventions

Activity	**Tasks**
Attitudes towards treating autism: Should we welcome genomic research to develop pharmacological treatments for autism? Why?	• *Two participants were asked to enact a short dialogue between two autistic people, one in favour and the other against the development of a hypothetical pill to cure autism:* Should we welcome genetic research to develop a drug to cure autism? Why? • *Participants were given a set of magnets displaying the following items with a brief explanation: genetics; neuroscience; treatments & interventions; services; societal issues.* Imagine that you have a set amount of money that you can spend on these research areas that are currently conducted on autism. As a group, please rank these on the whiteboard from the one you would spend the biggest amount of money on to the one you would spend the smallest amount of money.
Attitudes towards preventing autism prenatally: Should parents be allowed to use genomic information about their embryos' and foetuses' predisposition to autism to make reproductive choices? Why?	• *Participants read a letter in which the mother of two autistic sons discusses the potential development of PND for autism:* Do you agree or disagree with the mum's view? What may be the arguments in favour and against her view? Why? • *The same letter was used to investigate participants' attitudes towards PGD for autism:* Imagine that a couple who already has two children with autism is planning to have another baby. Their geneticist tells them that they can access a procedure by which they will be able to select an embryo with a lower risk for autism. Should this couple be allowed to do so to prevent giving birth to an autistic child? Why? How is this different from the mum's case?

Attitudes towards predicting autism to lessen its risk: Should we test babies for their genetic predisposition to ASD for early intervention purposes? Should we enrol high-risk children in early intervention programmes? Why?	• *Testing infants for their genetic predisposition to ASD:* Should we genetically test babies for this purpose? What might be the medical/psychological benefits and harms of knowing that a child is at risk of developing autism at a very early age, perhaps before they have shown any symptoms? Why? • *Enrolling infants in early intervention programmes:* Should we enrol babies in early intervention programmes before they have shown any symptoms of autism, on the basis that genetic test shows that they are at increased risk? Why?

The first activity investigated young people's attitudes towards *treating autism*. Groups discussed a thought experiment elicited by a dialogue between two autistic people, one in favour and the other against the development of a hypothetical pill to cure their autism, which two participants were asked to enact (see Appendix H). Although the dialogue was fictitious, I had grounded it on the opinions that two autistic advocates have publicly expressed on the topic (Higashida et al., 2013; Rubin, 2005). Because this was a thought experiment, it focused on the (unrealistic) option of a pill that eliminates autism altogether (a 'cure' for ASD); however, at the start of this activity I clarified that such a cure does not exist and I presented the dialogue as a hypothetical scenario. Moreover, prompts were asked to assess whether and how participants' arguments shifted if the hypothetical pill did not cure ASD altogether but treated some of its traits; on many occasions, participants spontaneously discussed alternatives to the proposed scenario. A subset of participants were involved in a ranking exercise about priority-setting in autism research (see Table 7).

In the second activity, young people were interrogated on their attitudes towards *preventing autism prenatally*. Groups commented on a letter that the mother of two autistic sons sent *The Guardian* in 2009, in which she reflected on the potential development of PND for autism (Moore, 2009). Participants were then asked whether and why their opinions would change if the scenario was about PGD. Both tasks were introduced by a brief recap of the difference between foetuses and

embryos, and of the different options to which PND and PGD could lead (i.e. termination of pregnancy and embryo selection).

Finally, the third activity investigated young people's attitudes towards *predicting autism* to intervene early and so lessen the risk of its development. First, I used the same letter described above to assess how participants' attitudes would change if testing was not performed before birth but early in life; young people were asked to focus on the medical and psychological benefits and harms of knowing that a child is at risk of developing autism at a very early age. The final task engaged the young people in a discussion about enrolling high-risk children in EIs, potentially before they have shown any sign of autism.

The activities in Part 2 investigated whether young people saw any normative difference between intervening at the different stages of life. During the discussions, I did not simply allow views and attitudes to emerge; to encourage participants to articulate on the fundamental motivations driving their attitudes I challenged participants' contribution with counterarguments and I asked comparative questions, which helped me explore how and why young people's responses changed when scenarios were not about autism but physical diseases or disabilities (e.g. 'Should we welcome genetic research to develop better treatment for cancer?', 'How would your response about PGD and PND change if the case was about deafness/paediatric cancer/heart disease?'). In this sense conceptual and normative analysis was to some extent integrated within data collection (Dunn et al., 2012).

Direct questions about biological citizenship and genetic responsibility were not asked in the FGDs, as this would have added another level of complexity to the discussion. Thus, the topic guides were entirely focused on autism. However, as I explain below, the analysis of the qualitative data was subsequently used to infer what the empirical data revealed about the meaning that young people attribute to biological citizenship and genetic responsibility in psychiatric settings; and the philosophical analysis of such responses served to draw normative conclusions on why and how

the responsibilities promoted by participants should underpin and guide action about the use of genomic information in psychiatric settings.

Immediately after each focus group, I listened to the recording of the discussion to compile field notes on how the discussion had proceeded. I particularly focused on whether certain activities needed to be reviewed or improved, as well as whether participants demonstrated particular interest in a certain topic. I then transcribed the discussion, which gave me an additional opportunity to take notes, this time on emerging themes. These notes allowed me to reflect on the progress of my data collection and inform subsequent focus groups; thus, data collection and analysis proceeded in an iterative way. I followed a similar process of note-taking to report on the main inferences that I drew between themes and categories emerging from the data during data analysis, which turned out to be useful during write-up, and as a tool for quality assessment of my methodology.

4.3.5. Data analysis

I transcribed five focus group recordings, which allowed me to familiarize myself with the data, while the remaining seven were transcribed by a transcription company. In the latter case, I verified all transcriptions by listening to the recordings and making edits if needed, which also helped me become familiar with the data (see Appendix J for the conventions adopted in the transcriptions). The Word documents of the transcripts were then imported in the qualitative research software NVivo 11 for data analysis. The most substantial analysis was undertaken between the two phases of data collection (September 2018), and at the end of the second phase (March–August 2019).

According to the tradition I align with, EB studies should aim at finding coherence between universal standards of justification of logically good arguments and relevant empirical and contextual data (Dunn et al., 2012). I achieved this through a process that integrated analysis of the empirical data and the philosophical analysis of such data. This process was informed by grounded theory (Corbin and Strauss, 2014) and constructivist grounded theory (Charmaz, 2014), and

adopted a coherence-seeking approach that draws upon the ‘grounded moral analysis’ methodology (Dunn et al., 2012) and the process of ‘analytic induction’ (Silverman, 2015).

While analysing the qualitative data, I first read my transcripts many times to ask exploratory and brainstorming questions of the data and get familiar with it. I took long memos to note down reflections that came to my mind, highlight arguments that I found unclear and make links between similar arguments across FGDs or between FGDs and the literature (Charmaz, 2014). When emerging categories were identified, I reflected upon relationships between them. In this process, I paid particular attention to group interactions (Belzile and Öberg, 2012; Rothwell, 2010). I took note of dominating voices and of voices of dissent; I highlighted passages when a participant’s argument made other group members change their mind or made the discussion take a new direction; I focused on specific terms that young people used to refer to autism and that were relevant to answering the secondary questions of my research, and I noted down differences in the use of such terms between individuals or across groups.

I then used a brainstorming approach to analyse my transcript and so develop codes that could be open to different interpretation of my data ('open coding', Corbin and Strauss, 2014). I coded my transcripts line by line and I initially tried to keep as close as possible to participants’ expressions and wordings to develop my codes (Charmaz, 2014). Because my topic guide was very structured, I initially coded activity by activity, i.e. I developed a coding scheme for each of the three activities in Part 1 and Part 2 of my FGDs. This allowed me to develop a very detailed descriptive coding scheme, in which I was primarily interested in highlighting what participants knew about autism (Part 1) and their arguments in favour/against the interventions in autism genomics (Part 2).

I then aggregated the open codes into higher-level codes ('conceptual coding', Corbin and Strauss, 2014). The coding scheme from Part 1 remained more descriptive because of the nature of the discussion and hence of the data, which were primarily focused on factual knowledge; in those activities that engaged participants’ normative thinking, the higher-level codes were given more

abstract names by using ethics language and terminology derived from the theoretical framework. In this way, in the higher-level coding scheme the set of core moral arguments, values and principles underpinning participants' attitudes were made explicit. I then used such set to inform subsequent focus groups, e.g. to refine my topic guide or identify new counterarguments to challenge my participants' views.

To progress in the production of the final set of claims that, when fitted together, generated a picture of young people's views of autism and moral attitudes towards using genomic information to predict, prevent and treat autism, the method of 'analytic induction' was used. This method combines the techniques of constant comparison and deviant-case analysis, which are used to validate study findings, as they test whether the emerging hypotheses are able to account for the data that are subsequently analysed, and fit together to constitute a coherent set (Silverman, 2015). This required me to compare data within and between transcripts and to keep going back and forth between data and analysis to test my emerging hypotheses (*constant comparative method*), as well as 'seeking out and addressing deviant cases [or data]' (*deviant-case analysis*) (Silverman, 2015, p. 99).

Following Dunn et al.'s EB methodology (2012) throughout data analysis participants' arguments were put under philosophical scrutiny, and challenged if they failed to meet the universal standards of reasoning in moral argumentation, hence continuing the conceptual analysis initiated within the FGDs. In particular, invalid arguments were discarded if it proved too difficult to understand what participants were trying to say, but only after a thorough comparison within and between FGDs that failed to unveil a possible understanding; when participants neglected contingent features of the real world (and so their arguments were valid but unsound), data were interrogated to understand why this was the case. As I will show in the result chapters, this occurred when participants neglected the complexities that arise at the intersection between genomics and psychiatry, by overemphasizing autistics' special talents or the causal role of genes in the

development of ASD. In these cases, they promoted stereotypical views about autism or deterministic views, which do not fit the current understanding of autism and ASD genomics.

In this way, young people's insights were used to contribute to developing a normative understanding of how they thought genomic information about autism should be used; however, their attitudes and arguments underpinning such attitudes were also subject to normative criticism to identify where young people might have proposed invalid or unsound arguments. This was essential in order not to undertake a form of 'ethics by opinion poll', whereby empirical data (e.g. participants' opinions about how autism genomic information should be used), is taken as a direct source for justifying how people should behave within a certain practice (Salloch et al., 2014).

In line with Dunn et al.'s 'grounded moral analysis' (2012), I reiterated the above-described process until I was able to shape a range of conclusions that could be justified because they were the result of a process of analysis that produced coherence between universal standards of logically sound arguments and empirical data on young people's views and attitudes. This was, however, not meant to achieve 'hard' normative work, but to elucidate the moral concerns that young people raise about autism and ASD genomic advances, which could in turn contribute to answering the overarching primary research questions. The final coding structure is shown in Appendix K (for FGD Part 1) and Appendix L (for FGD Part 2), around which the next two chapters are structured.

In this research, I could not achieve inter-coder reliability in a systematic way due to the lack of colleagues with enough expertise on the research topic and enough time to meaningfully engage with the coding process. However, in the early stages of data analysis I met with colleagues from the NEUROSEC team and the Ethox centre, who were asked to read one study transcript in advance, and note down the main themes they identified and any arguments they found surprising or worthy of attention. This meeting confirmed some of the emerging hypotheses that I had developed (e.g. young people's dimensional understanding of autism, see Chapter 5), hence

reducing the risk of potential biases; and it encouraged me to give more relevance in my analysis to the way in which participants reasoned around the research topic.

Moreover, during data analysis I had regular meetings with my supervisors to discuss, in the early phases, the long memos I wrote when analysing each FGD and, subsequently, my emerging categories, which helped me refine the labels used to refer to participants' arguments and to progress in the development of my evolving coding structure. Further, as suggested by Riessman (2007), I kept a diary of the critical decisions I made to inform subsequent stages of my data collection and of the inferences made in the development of the coding scheme during data analysis. This was meant to increase critical self-awareness and assess the impact that decisions and inferences made on the development of my project and its normative conclusions (Silverman, 2015).

Having addressed the theoretical underpinnings and the design of the present study, to further prove my commitment to rigorous argumentation in the next section I will undertake an exercise of scrutiny over how my intuitions and experiences may have influenced the design and conclusions of my study (Seale 2004). Specific methodological limitations, and the extent to which they might have introduced biases in the data, will be discussed in Chapter 7.

4.4. Reflexivity

In the context of the present study, reflexivity, i.e. 'a critical interrogation of the relationship between the researcher, the world she is studying, and her experience and awareness of that world' (Ives and Dunn, 2010, p. 261), raises questions about the impact of my work with the NeurOx YPAG on the research process. On the one hand, I believe that this experience has been extremely useful for the success of my FGDs. Indeed, I had developed a certain conception and appreciation of young people's capacities to contribute to ethics debates, which resulted in my genuine interest in their contributions. This is likely to have become manifest to my research participants, who may

have felt encouraged to engage in the discussion by seeing that I was taking them seriously. Moreover, I had gained an understanding of methods, tools and ways to elicit their thoughts and contribution to ethics debates, which may have improved the quality of my dataset.

For instance, my familiarity with working with young people may have helped reduce the power imbalance between me and my participants so that they did not feel like I was testing them. This was particularly important not only because some young people find discussing genetic concepts complicated (s4.3.4.2), but also because, out of respect to researchers, younger children tend to confirm what they are told (Lewis, 2002; Singh and Keenan, 2010). To overcome this issue, I made the FGDs as little academic as possible (with an awareness that I had a responsibility to address some misconceptions about autism), and I encouraged participants to reflect on both arguments in favour and against a certain issue, before asking them to express their own opinion. Having noticed that, regardless of participants' age, some groups tended to be quieter, I also introduced peer-led questions as a research tool, whereby participants asked the first and main question of each activity described in s4.3.4.2, drawn from a pile of flashcards, and I only interjected when follow-up or prompts were needed (Pavarini et al., 2019).

On the other hand, I was convinced of the importance that my intuitions regarding young people's capacities were grounded on robust argumentation to avoid that the enthusiasm towards children's involvement in research led me to read too much into their accounts – the 'epistemic privilege' (Krimerman, 2001, p. 70), whereby participants' accounts are uncritically privileged over researchers' analysis and interpretation (Holland et al., 2010). These considerations encouraged me to develop a justification for minors' involvement in this study, which I have proposed in s2.4.

In contrast to my extensive experience with youth, when I started this research I had little direct familiarity with ASD and autistic people. Scholars have discussed the (dis)advantages of (not) belonging to their study populations in qualitative research (see Dwyer and Buckle, 2009), which relates to my position as an outsider to the autistic and autism community. This distance from the

reality of autism helped me to be objective when discussing ethical issues around ASD genomics; and it contributed to reduce the researchers–researched power imbalance, as it allowed me to defer to study participants with relatives on the spectrum as a source of expertise on the research topic. However, after the first FGDs I realized that while I had an in-depth academic and philosophical knowledge of and interest in autism, I lacked experiential knowledge of it, which some consider as essential for empirical bioethicists (Ives, 2008; Singh and Keenan, 2010).

Therefore, even though I used autism only as a case study, I felt that I needed to develop some form of engagement with people on the spectrum. This is why after the first phase of data collection I sought opportunities to volunteer with autistic people, which I found in the local charity Autism Family Support Oxfordshire, by supporting staff members at the youth groups that the charity runs for 13–16-year-old young people with high-functioning autism. For logistical reasons I could only be enrolled in the team of volunteers in July 2019, when I had already completed data collection. However, this experience has made me feel more legitimized to talk about autism, and has given me an increased sense of confidence in my capacity to master the research topic, which helped in the data analysis and writing-up phases.

4.5. Concluding remarks

This chapter has outlined the methodological choices underpinning the empirical study that I conducted to answer the primary questions of the present research, by using autism as a case study. I explained that I adopted a qualitative methodology in order to achieve a deeper understanding of young people's views of autism and attitudes towards using genomic information for the prediction, prevention and treatment of autism. To this purpose, I conducted focus group discussions with a general population of young people between 13 and 18 years old, who were recruited via schools and youth groups in Oxfordshire, London and Birmingham. The design of this empirical study was informed by a consultation with the NeurOx YPAG, in line with a co-production model of research.

Focus groups provided the empirical data to address a range of secondary questions that could contribute to answering the primary questions of the present research. In the next two chapters, I will present the results of the analysis of the qualitative data and of the philosophical and conceptual analysis of participants' responses, to offer a picture of young people's views of autism (Chapter 5) and moral attitudes towards using genomic information to prevent autism at different life stages (Chapter 6). In Chapter 7, I will discuss what young people's arguments tell about the meaning they attribute to 'biological citizenship' in mental health settings, and what responsibilities young people promote under their framework of biological citizenship. As required by a study that follows the tradition of empirical bioethics, I will draw normative conclusions about why and how such responsibilities should underpin and guide action in the practical context at the intersection between genomics and mental health.

Chapter 5. Young people's views of ASD: Dimensionality and uncertain nature

5.1. Introduction

This chapter seeks to present the results of Part 1 of the focus groups on young people's views of autism, and to shed light on the dynamics through which a certain account of autism is developed in groups where most people have no special relation to the condition. Because different models of autism exist (Chapter 3), at the start of the empirical phase of this research I hypothesized that participants' desire (or lack thereof) for the application of genomics to the primary and secondary prevention of autism may be influenced by the way in which they conceptualize the nature of ASD. Thus, this part of the FGDs primarily aimed at investigating where young people positioned themselves in the debate about the three models of autism.

However, because of the involvement of lay participants I deemed it important that such an investigation followed a discussion about young people's understanding of ASD traits and aetiology to help participants get to a level of engagement with the topic to feel comfortable about discussing how they thought autism should be conceptualized. Therefore, I developed three secondary questions, which I will address in this chapter:

1. What do young people understand autism is?
2. What do young people think autism is caused by?
3. Do young people consider autism as a disorder, a disability, or a difference (and why)?

5.2. Overview of the present chapter

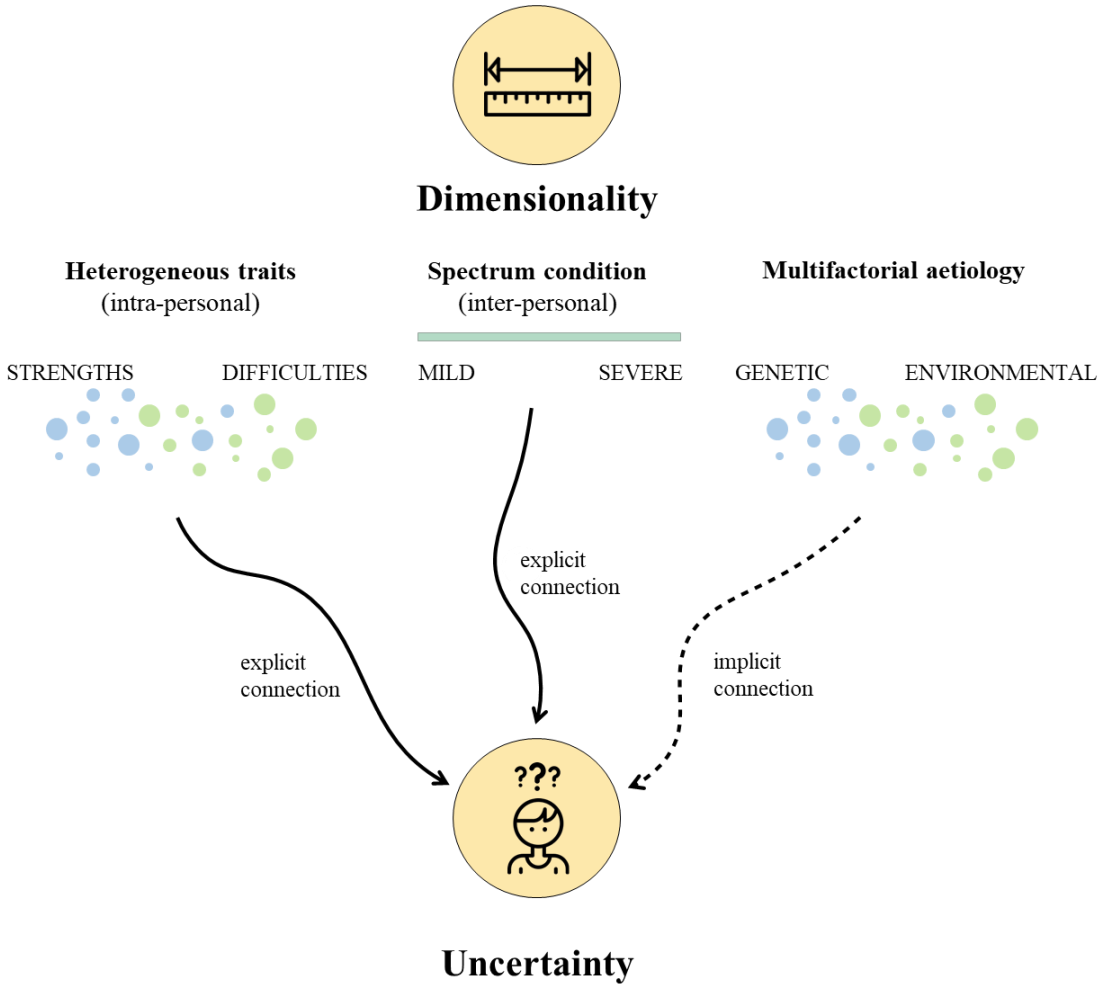


Figure 7: Young people's views of autism

Data analysis identified two main categories in Part 1, i.e. 'dimensionality' and 'uncertainty' (Figure 7). As I will present in sections 5.3 to 5.5, young people conceptualized autism as a dimensional condition in three ways. They described ASD in terms of heterogeneous behaviours, from clear difficulties, to clear strengths, to traits that can be at times strengths and at times

weaknesses; they were aware that autism is on a spectrum from mild to severe forms; and they developed a multifactorial account of ASD aetiology as aligned on a dimension of genetic to environmental factors. Thus, although I asked dichotomous questions (strengths vs difficulties, mild vs severe, genetics vs environment), participants' conceptualization of ASD on a continuum of traits, severities and causes highlighted the unfitness of dichotomies to the study of autism.

Young people used their dimensional understanding of autistic traits and severities to explain their uncertainty about the nature of autism (s5.6). No strong belief was indeed expressed about whether ASD should be categorized as a disorder, a disability, or a difference, but rather a variety of opinions were proposed. Interestingly, young people did not discuss the nature of autism in light of their considerations about ASD aetiology and their preference for a biological understanding of ASD. However, the dimensionality of ASD aetiology implicitly contributed to create a complex account of autism that fed into young people's uncertainty about how to conceptualize the condition.

Throughout this chapter, I will show that the philosophical scrutiny of young people's arguments identified a tension whereby at times young people abandoned their dimensional account of autism and uncertainty about the ASD nature to adopt more 'comfortable' dichotomous and clear-cut views that could support certain narratives about the condition. For instance, participants sometimes placed undue emphasis on autistic strengths to develop a positive conceptualization of autism; or on the causal role of genes to back up certain intuitions that they had about the way in which autism develops.

On the one hand, the 'strategic' selection of certain bits of knowledge is a cognitive mechanism that helps people navigate the discussion of complex phenomena, and so meet certain context-dependent goals; in this sense, tensions between dimensional and dichotomous thinking should be embraced rather than discredited in favour of some academic ideal of coherence (Condit, 2019). On the other hand, when positive autistic traits were accentuated young people ended up developing

stereotypical views about autism, hence promoting myths about autistic special talents; when they emphasized the genetic bases of autism, deterministic assumptions about ASD aetiology were proposed, which may lead to misinformed choices about ASD genomics. Therefore, as I will further suggest in the concluding chapters, there seems to be value in promoting dimensional thinking and uncertainty in the complicated context of psychiatric genomics.

5.3. What is autism? Heterogeneous traits across a spectrum

Overall, participants appeared to be quite knowledgeable about autistic traits. As source of evidence of their knowledge they used the behaviours that their autistic friends or relatives show or what they had learned about autism from books, movies, or TV series. The analysis of this part of the focus groups identified two subcategories, i.e. 'heterogeneity' (s5.3.1) and 'spectrum' (s5.3.2).

'Heterogeneity' is often associated with autism; indeed, nowadays many scholars refer to ASD in the plural form ('autisms', see Coleman and Gillberg, 2012; Singh, 2016b). However, the term is attributed various meanings (Fitzgerald, 2012). Although participants did not explicitly call autism 'heterogeneous', in this research this category refers to three aspects of their understanding of ASD. Young people thought that, within the same individual, autism presents a number of different traits; they understood some of these as positive and some as negative, hence moving beyond a purely medical model focused on deficits; and they argued that some behaviours are advantageous or disadvantageous depending on the context and the extent to which they are manifested. The idea that positive and negative traits lack a clear-cut distinction contributes to create a picture of autism as a dimensional condition, and resonates with the views that autistic adults expressed about their own behaviours in a recent qualitative study (Russell et al., 2019; see s3.4.3).

Participants also showed that they were familiar with the idea that, across different individuals, autism presents a 'spectrum' of mild to severe forms, and they often spontaneously used terms such as 'spectrum', 'scale', or 'range' to describe autism. In the next pages, I will refer back to the

following passage from FGD4, in which Mia, Evelyne, Jasmine, and Amelia discuss their understanding of ASD traits (Box 6).[26]

Box 6: Young people's familiarity with ASD heterogeneity and spectrum
1I:[27] I want to start with a very generic brainstorming on autism. So what comes to your mind when you think of autism? Do you know what autism is? 3M: Kind of… I don't know, I think **they have difficulties like understanding people's emotions**.[28] 5I: mmm… 6M: And kind of, like, **interacting with people.** 7I: Yeah … yeah, what else? 8E: **Sensory issues**, I think as well. 9I: mmm… What do you mean? 10E: So, like, if there's lots of noise or lots of stuff going on at the same time they can get kind of overloaded with it. 12I: mmm… 13J: Yeah and **they like very specific structure and very logical routines** 14I: Routines, yeah. Can you give an example, like… do you know…? 15J: hmm, like my friend always has to have lunch at the same time every day. 16I: ok yeah, yeah what else? Do you know anything else? Difficulties or maybe strengths? 17A: They find it difficult to learn a language, like I don't know how to explain it… […] 19J: I think **it depends on how high functioning you are**. So really high-functioning people can have a specific talent like in maths or science in which they really excel on, and then on the other end of the **spectrum** there are people, how do you say, can't… haven't learnt how to speak because… it varies. 23I: Yeah so maybe they have learning disabilities. 24M: And I think they [autistic people] become **very obsessive with certain things,** like, if they... I don't know, like, I know someone who is just, I don't know, they are obsessive about dinosaurs, when they were young they were really, really obsessed with them… I don't know. 27I: And do you think that's a strength or a difficulty? 28M: mmm… I think it can be both sometimes? 29I: Why do you think so? 30M: hmm… oh, I don't… I don't know I think because, like, I know that he would all read about them and would know all about them and I don't know… but then he would also kind of just… not read or watch other movies and stuff. He would kind of just only focus on that. 33I: Yeah, I understand. 34M: I don't know if it makes any sense. 35E: Because if you know a massive load about something you are very good at, **obviously that is a strength**, but also if you are very focused on it **you may not be talking about other things** and other people don't really always want to listen to you talking about dinosaurs or whatever. 38*A, J, M laugh* 39I: So I guess that links back to having difficulties socializing. 40E: Yeah.

[26] In Chapters 5 and 6, FGD exchanges in boxes are representative of similar passages recorded across FGDs.
[27] In all FGD exchanges in boxes, 'I' stands for 'Interviewer'
[28] In all FGD quotes in Chapters 5 and 6, I highlighted bits in bold when they are relevant to the analysis.

41J: **Or it could affect your school work** if you don't wanna learn how to study a book if you are only interested in dinosaurs or something and, like, in the future you have bad grades and then…
[…]
45I: I wanted to show you a quote by Stephen Shore who is a professor who has autism who once said… you may have heard of it because it's quite a famous quote, when he said that 'If you have met one individual with autism, you have met one individual with autism'. So what do you think that means?
49J: **That they are all different**, like, you can't just assume because you know one person with autism they all have the same traits and stuff.
51I: yeah yeah what else? Do you know that autism is also called autism spectrum disorder? Do you know what 'spectrum' refers to?
53A: Everyone is somewhere on the spectrum so…
54I: Yeah, what do you mean?
55A: hmm… like Jasmine was saying **they all have, like, different traits** and they could have, you could start talking to that person and you could immediately see that they have autism so **they got really strong traits of it, but then you could speak to another person and only when they tell you that they have autism you didn't know because you couldn't see any traits**.
59I: Yeah, do you want to add anything else? No?
60M: hmm… I don't know, just kind of, **you can have, like, an extreme and then someone in the middle and then yeah**…

5.3.1. Difficulties, strengths, or both? ASD heterogeneity

Data analysis showed that young people associate a range of behaviours with autism (Figure 8).

This is therefore the first meaning of the sub-category of heterogeneity.

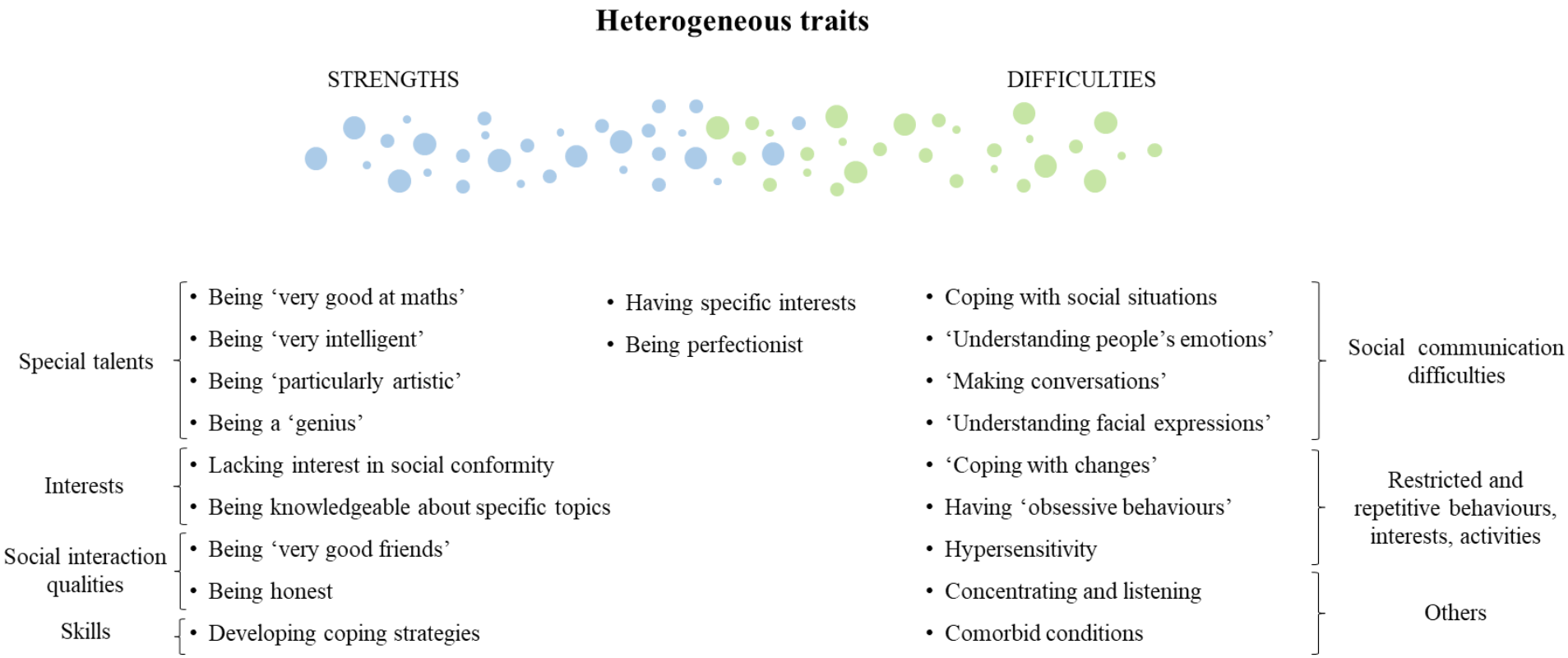


Figure 8: Most frequently mentioned ASD strengths and difficulties

When asked to brainstorm about autistic traits, in most groups participants first thought of difficulties (see Box 6), and only when prompted did they describe positive traits, which may suggest that they initially held a deficit model of autism aligned to the medical understanding of the condition. This is confirmed by the kind of language that participants used at the beginning of the FGDs, when they described autism in terms of having 'difficulties', or a 'problem'. In the quote below Laura uses such a terminology, even though she is unable to provide any concrete examples of difficulties that autistic people experience. Brainstorming activities are interesting in this sense because they expose people's preconceptions:

> Laura: **Difficulties**, like, in life and stuff.
> Interviewer: In what, sorry?
> Laura: In life. Like, doing stuff, they might have **problems** and that.
> Interviewer: Can you give an example?
> Laura: Um, I don't know. (FGD6)

Participants listed several difficulties, among which the most frequently mentioned fall under the two sets of impairments that the DSM-5 attributes to autism (Figure 8). Past empirical studies on lay understanding of autism have reported that social communication difficulties are the challenges that people most commonly associate with autism (Jensen et al., 2016; John et al., 2018; Mitchell and Locke, 2015). For instance, in Box 6 Mia mentions difficulties in 'understanding people's emotions' (lines 3–4) and 'interacting with people' (line 6). Among repetitive behaviours young people listed difficulties in coping with changes (liking a 'very specific structure' and 'very logical routines', line 13 in Box 6); and having 'obsessive' behaviours, i.e. getting fixated on niche topics (see Mia's comment in lines 24–26). Like Evelyne in lines 8–11, some participants also referred to 'sensory overload' or hypersensitivity. Other frequently mentioned issues were comorbid conditions such as ADHD and anxiety, and difficulties in concentrating and listening, which participants often observed in school contexts:

> Scarlett: Two children I have met once both had quite severe autism. And **they were both really easily distracted**. They would mess about, play about, walk about, talk to anyone. And in the second that they'll say, 'right, you have to do this test now', they'd be done so quickly. But they were still very easily distracted. But then when they actually could sit straight they quickly got work done so easily. It just stuck in their mind the whole time, it was still in the back of their mind but **they just saw so many other things that would distract them**. (FGD11)

As this quote shows, in young people's views, ASD provides people with strengths too ('they quickly got work done so easily'; 'it just stuck in their mind the whole time'). This demonstrates the second form in which the sub-category of 'heterogeneity' emerges from the data, i.e. that autistic individuals present a variety of both negative and positive traits. When prompted to discuss ASD aspects that they saw as positive, most groups tended to think of special talents, such as being 'very good at maths', 'very intelligent', a 'genius' and 'particularly artistic' (Figure 8).

Special talents were not the only positive traits that groups identified, which once again demonstrates young people's familiarity with the variety of behaviours that autistic people show. They also mentioned that autistics tend to be less receptive to social norms and the need to conform to social standards. Autistics were also seen as resourceful because they tend to develop coping mechanisms to deal with their difficulties and, despite struggling in social situations, they have personal qualities (e.g. being good friends, being honest) that participants valued in social relations:

> Alice: And I think once you do form a relationship or a connection with someone *[with autism]* **they'll be a very good friend**, or, like, a good person. It just takes a bit longer to develop that relationship. (FGD3)

Finally, the young people seemed to think of some traits as on a continuum from aspects that are positive to aspects that are negative to blurred traits. As exemplified by Mia's comments about 'things' autistics become 'obsessive' about (Box 6, line 24), on some occasions participants presented autistic behaviours in a way that made it unclear whether they understood them as positive or negative. Upon my request for clarification, participants argued that the same trait could

be seen as both a strength and a difficulty, and data analysis showed that this depended on the extent to which autistics show their behaviours, and on the social context.

In Box 6 above, Evelyne suggests that if somebody is very passionate about a topic and becomes very knowledgeable about it, 'obviously that is a strength'; however, if the person's interest is so specific to the extent that they are unable to engage in conversations about other topics, that could become a difficulty in social relationships. To this argument Jasmine adds that specific interests could also become a problem in specific contexts, such as school, if they prevent people from learning about other topics (lines 41–43). In this way, the same trait could range from being understood negatively as an 'obsessive behaviour', to being thought of neutrally as a specific interest, to being a positive trait that makes people very knowledgeable about the topic they are passionate about (Figure 8).

Within a similar discussion in a different focus group, Ludovica suggested that some autistic traits might become difficulties due to the way in which society works. This shows that when co-constructing their account of autism, young people shifted away from their initial endorsement of a narrow deficit model of autism to acknowledge broader societal and structural determinants of whether certain ASD traits are perceived under a positive or negative lens:

> Ludovica: I guess **that can be a difficulty in terms of the way that our society works** because they are kind of expected to be able to do more than one thing, but also if you can get into a job where you only have to focus on the one thing that you are interested and passionate about it can be a real strength. (FGD1)

This third way in which 'heterogeneity' emerges from the data confirms that, despite my dichotomous questions about autistic strengths and difficulties, young people understood autism as a 'fluid' condition that causes traits on a dimension from advantageous to disadvantageous behaviours. In other words, heterogeneity was thought of in a dimensional way.

5.3.2. Autism as a spectrum condition

Not only did participants think of autism as causing both positive traits and challenges, which in their understanding have blurred boundaries. They were also familiar with the idea that autism manifests across a spectrum of different severities that range from mild to severe (Figure 9).

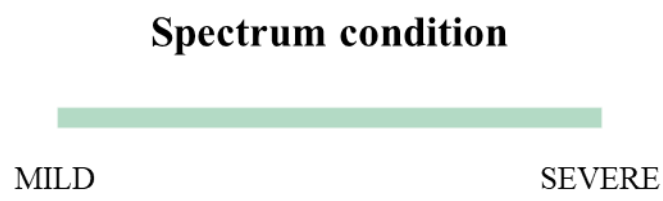


Figure 9: The spectrum nature of autism

Young people often introduced the concept of the autism spectrum spontaneously, and on some occasions participants explicitly used the word 'spectrum' (see Jasmine in Box 6, line 21), or similar terms (e.g. 'scale' or 'range'). Groups who did not initiate a discussion about the autism spectrum indicated that they knew about it when prompted to comment on the quote, attributed to Professor Shore from Adelphi University, 'If you have met one person with autism, you have met one person with autism'. Some young people seemed to assume that the spectrum nature of autism is common knowledge ('obviously'):

> Jason: My understanding of it *[Asperger syndrome]* is, I would say what I understand of it is it's a slightly more specific area, so more specific symptoms. **Whereas with autism obviously it can be any different range of symptoms.** Asperger's is sort of more like a set few, I'd say and it is stuff like struggling to process, social interaction, so struggling with that. That's what I know it is. (FGD10)

While 'heterogeneity' is an *intra*-personal quality, the sub-category of 'spectrum' refers to the different severities that autism can have across individuals and is therefore an *inter*-personal quality. For instance, in addition to participants' description of the various traits that they associate

with autism, in Box 6 (lines 56–58) Amelia argues that there are some autistics 'you could immediately see that they have autism so they got really strong traits', while with others 'only when they tell you that they have autism you didn't know because you couldn't see any traits'; Mia adds that 'you can have, like, an extreme and then someone in the middle'. Both subcategories suggest that young people had a dimensional understanding of autism, which contributed to their uncertainty about the nature of the condition, as I will discuss in s5.6.

Given participants' awareness that some people have severe autism and their familiarity with the ASD spectrum, it is surprising that most of them associated autism with special talents, as presented above. These traits, which in the literature are called 'savant skills' (Draaisma, 2009), can be identified only in a small proportion of people on the spectrum, yet they are among the misconceptions that lay people tend to have about autism (see John et al., 2018). According to autistic people and their families, this creates high expectations of autistics to have special talents, and draws (self) stigma on those who do not show them.

In this research, only a minority of groups mentioned that the idea of autistics' savantism is a stereotype, as shown in the quote below:

> Marta: […] Um, yeah, I do think there is a kind of **stereotype** as well, isn't there, you know, really good at maths or something like that?
>
> Simon: Oh yeah, yeah.
>
> Marta: I don't know, **I don't know how true that is** *[laughs]*. (FGD9)

In most FGDs, unless challenged participants tended to assume that all autistic people are very skilled in certain tasks, or used a language that suggested so. According to some authors, this misconception is due to misleading media representations of ASD (Draaisma, 2009). This may apply to this research too, given that, as mentioned, movies and TV series are among the sources that young people used to develop their understanding of ASD. However, there may be two alternative explanations to the idea that young people are uncritical and passive recipients of media

representations. First, participants reflected on their autistic friends, schoolmates, or relatives' behaviours to make claims about autism, and young people may engage more frequently with peers with milder forms of ASD, who tend to be more integrated in society and in mainstream school. Among these it is more likely that they will know whether someone has autism if they also show special skills.

Second, in the past years some scholars have criticized the late 90s assumptions about public understanding of science, genetics and health conditions, which had adopted a deficit model to support the importance of educating the publics around increasingly pervading scientific notions and technologies. This scholarship, which is relevant to this thesis's critical take on the concept of biological citizenship, has unveiled the complex ways in which beliefs, attitudes and discourse function, which the deficit model had overlooked. It showed that the lay publics hold sophisticated understandings and views that are grounded on pre-existing knowledge and particular social and cultural backgrounds (Marks, 2016). In a recent paper about lay people's (alleged) genetic essentialism that falls under this tradition, Condit argued that when reasoning around and making choices about heterogeneous and complicated phenomena such as health, lay people 'tend to store multiple categories […] that they deploy "strategically" to serve context-dependent goals in light of varying combinations of immediate and long-term interests' (2019, p. S27). In this sense, lay people should be seen as 'strategic' rather than 'genetic' essentialists or determinists, as had been for long postulated by social scientists.[29]

As the discussion about the nature of autism further demonstrates (s5.6), the philosophical analysis of participants' views of autism and their underpinning arguments showed that, although in my research young people had a dimensional understanding of ASD, at times they seemed to

[29] In this literature, the terms 'genetic determinism' and 'genetic essentialism' are often used interchangeably. However, in the present thesis the former refers to the belief that genes are the sufficient cause of species, group, or individual characteristics; the latter refers to the tendency to assume certain characteristics of a person or groups because of their genetic make-up (Wachbroit, 2000).

'strategically' emphasize its positive aspects to distance themselves from negative accounts of the condition. In this way, they moved from the 'unstable' position of dimensionality and uncertainty, to a more 'comfortable' position that celebrates one side of the dimensionality only (strengths). However, in so doing they made comments at odds with the idea of the ASD spectrum and developed stereotypical views that grant autism the nature of 'difference' only because 'autistic people have special talents'. As I will show in the next section, this argumentative strategy was also deployed when young people discussed risk factors for autism.

5.4. Genetics, environment, or both? ASD has a multifactorial aetiology

'Multifactorial aetiology' is the third sub-category to describe young people's views of autism. In most cases participants attributed a higher degree of causation to genetics, hence describing autism as a biological condition, which resonates with other studies exploring lay beliefs about ASD aetiology (see Furnham and Buck, 2003; Mitchell and Locke, 2015; Park et al., 2018). However, the group setting allowed for strong biological accounts to be mitigated by those participants who argued that environmental factors play a role in the development and severity of ASD, and so groups eventually identified multiple causes of autism as aligned on a continuum from genetics to the environment.

Thus, this section will show that, despite having a different expertise from scientists' 'privileged' knowledge, in their negotiation of different beliefs young people co-constructed an account of risk factors for autism that embraces the complexity of ASD aetiology. To support their beliefs they relied on evidence drawn from the experiences of autistic people they knew; biology classes; or science news they had heard of or read about. For instance, differently from studies conducted with older people (Mitchell and Locke, 2015), most groups were aware of the Wakefield MMR vaccine

scandal (s3.2), which is promising news given the publics' ongoing scepticism about vaccines. In this study, irony was often used when participants discussed the topic of vaccines:

> David: What do you think causes autism and why?
> Luca: Vaccines. *[laughing]*
> All: *[laughing]*
> Interviewer: Is that for real or are you joking?
> Luca: I'm joking, I'm joking. (FGD12)

Participants' negotiation of a more biological and a more environmental account of autism is illustrated by the following passage from FGD11, in which Leo, Denise, Ruby, Evie and Scarlett discuss their understanding of ASD aetiology (Box 7). This exchange shows that the young people ultimately questioned the strong genetic account of autism supported by Scarlett, and concluded that some environmental factors must contribute to autism.

Box 7: Young people's negotiations about ASD multifactorial aetiology
1L: Well, **genetics affects the structure of your brain. And I would say the structure of the brain can lead to many disabilities**. I would put it a lot more towards *[genetics]*. *[Leo refers to the magnet in the visual analogue scale, see s4.3.4.2]* 4D: Yeah, I would put it more up here *[towards genetics]*. 5R: You can also say that **some women who smoke during their pregnancy** cause children to be underdeveloped as they grow up. 7E: Yes. 8D: Yes. 9S: **It's hard because loads of women may smoke.** They say you can't do loads of things when you're pregnant. You wouldn't be able to eat sushi. You can't eat rare steaks. You can't eat soft cheeses. 12R: You can't eat sushi? *[In a shocked tone]* 13 *Many laugh* 14S: There are things that may affect the development of your child. Women used to smoke and be pregnant all the time. That was the norm and loads of babies come out perfectly healthy and fine. 17L: **Well, would you argue, if it's from a while ago, we may not have had the same tools to diagnose the illnesses even though they were present?** 19R: Maybe it's just people who are drug addicts that are always [unclear] as well. 20S: It's just hard to say because I have even known women who smoked for their pregnancies and there is nothing **wrong** with their baby *at all*. **I think it's *definitely* more genetic because if you come out with your brain wired differently,** you're automatically going to be different anyway, no matter what. You can't, I don't feel like you can necessarily… Maybe, yes, if your baby comes out slightly early than others because it's a starving of oxygen. If the baby suffers from starvation of oxygen from the brain.

26R: Yes, it could be a [unclear] of things.
27S: And it is going to affect the way you feel because that is a mental disability. **But it's definitely, I think, genetics.**
29L: Yeah.
30R: I think it is, because if you think about it some people can diagnose their children while the baby is still in the womb. You can have blood tests done to see if your child will have a certain disability.
33S: Yes, like Down's syndrome. You can find that out early now. I think it's definitely more genetic than it is environmental.
[…]
36S: **Of course** because if a child comes out perfectly normal, the birth is fine, the child comes out to date, I mean nine months after pregnancy, and there wasn't a lack of oxygen in the birth, the birth is all fine, the child still comes out with a mental disability then **it's *clearly* only to do with genetics.**
[…]
41E: I think environmental factors can cause autism though because in EPQ *[Extended Project Qualification]* I'm researching if parents can cause kids to become criminals. And I researched this person called Rene. She found out there's certain ways a parent can raise a child and they can have more mental health issues.
45R: Yeah.
46E: Although you can still have that kind of parenting and your kids not have those mental issues, **I think it is definitely a cause that can cause autism.**
48S: Do you think you develop autism or do you think you're just born with it?
49E: I think you can develop it in some ways but you can also be born with it.
50 Others: Yeah.
51E: It's just to do with parenting and your environment.
52R: I think as **people low on the spectrum kind of develop a bit more as they grow up and how the parents are** [unclear]. But I think if you're very high up on the spectrum I think it is more genetic.

5.4.1. Leaning towards a biological account

Most groups endorsed a neuro-genetic account of autism, which is confirmed by the language that they used throughout the FGDs, in which they described autism in terms of genes, brain chemistry, or issues that happen during children's brain development:

> Caroline: I find it hard to imagine why environmental factors may contribute because it is so scientific, **autism is a condition that is so much placed in the brain that I do not see how the environment may play a role** […] For example, in cases where people with autism are unusually intelligent, **it's very hard to believe that this was caused by an environmental factor rather than being 'pre-programmed'**. It seems, in my opinion, that autism is **too linked with the brain** to be a result of anything other than this! (FGD3)

Four main arguments were provided by participants to support their neuro-genetic account of ASD aetiology. As Leo initially argues in Box 7, autism is linked to brain development, and genes affect the way in which the brain develops. Participants also argued that people cannot *become* autistic; rather, autism is a congenital condition. This is the argument Scarlett seems to be alluding to in Box 7 (line 48), when she challenges Evie's point about the role that parenting might play in the development of autism. From this assumption young people concluded that autism must be caused primarily by people's genetic makeup, which makes the person susceptible to autism:

> Camila: Because you can't just, like, **I don't think you can just, like, turn autistic** which would be if your environment; **it has something to do with your brain which is, like, genetics**. (FGD5)

Moreover, as documented in past studies, lay people build on their pre-existing forms of knowledge to develop new understandings of health, science and technologies (Marks, 2016). In this research, some participants grounded their belief about the genetic underpinnings of autism on the genetics they learned in biology classes:

> Greta: Yes, I agree. It's probably that you have genetics combined in the mother and father when the child is made. Maybe there's some gene thing that doesn't go together and just messes things up.
>
> Interviewer: Why do you think so? Have you read it somewhere or is it based on something that someone else has told you or just thinking of people that you know who have autism. []
>
> Greta: I don't know. It's basically just, like, on **previous knowledge of what we've learned in biology about genes and stuff**. Because that could be a reason why. (FGD8)

Furthermore, those young people who knew of families in which several members are on the spectrum spontaneously discussed whether autism might be passed down genetically:

> Gracie: I'm not entirely sure, because, like, **my dad had it and then my brother had it, so it could be genetics.** But my brother was also born early as well, so that could also have had something to do with it, so I'm not entirely sure which way to go *[either more towards genetics or environment]*. (FGD7)

Within this discussion, young people's endorsement of the complexity of ASD aetiology emerged in the sense that all but one group argued that autism is caused by multiple genes rather than one gene, for which participants provided two main reasons. The first pragmatic consideration was that scientists would have identified the 'autistic gene' by now, if it existed. Second, if it was caused by one gene only, that gene would be expressed in the same way in everyone, and so all people with autism would have the same symptoms; however, this would be at odds with participants' awareness of the spectrum nature of autism. Although this argument may not align with the current understanding of the variable expressivity of genes, it shows that young people were capable of articulating sophisticated reasons behind their views, hence providing further evidence of the limitations of a deficit model of lay expertise in genetics:

> Caroline: **I feel, like, they would have found it by now if there was one gene, with the technology we have** I don't think that…
>
> Alice: And then you would be able to tester it.
>
> Interviewer: To what, sorry?
>
> Alice: Like, if there was just one gene, you would be able to tester it, then you wouldn't… **it'd be harder to put people on a scale**.
>
> Caroline: Y**eah, on the spectrum**.
>
> Alice: **Or on a spectrum, if there was just one gene**.
>
> Caroline: It would be you do or you don't.
>
> All: Yeah.
>
> Sophie: You definitely have it or you definitely don't because **gene expression would be the same**.
>
> Caroline and Alice: Yeah. (FGD3)

The complexity of young people's account of ASD aetiology is also confirmed by the evidence that they ultimately developed a multifactorial understanding of the condition, to which I now turn.

5.4.2. But environmental factors are also acknowledged

Despite most groups arguing that autism is primarily caused by genetic factors, strong conceptualizations of autism as a biological condition did not go uncontested, as the interaction in

Box 7 showed. On the contrary, environmental factors were acknowledged as impacting on both the development of autism and its severity. In this way, by negotiating different beliefs young people developed a multifactorial representation of autism aetiology lying on a continuum from genetics to the environment (Figure 10).

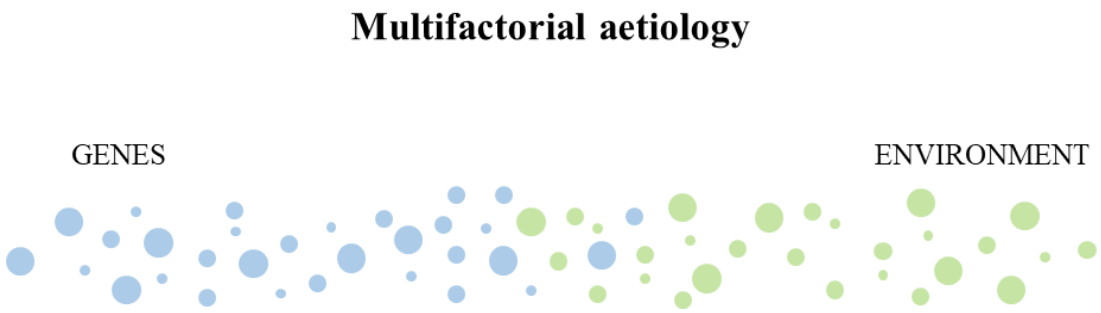


Figure 10: ASD causes range from genes to the environment

Since participants linked autism to people's genetic makeup and brain development, to reconcile biological and environmental explanations they often reflected on how the environment could impact on genes and brains. Three main arguments were provided. As aforementioned, from the idea that autism is a congenital condition, participants described autism as having to do with people's biology. However, they argued that the environment can impact on this biological *substratum* by making symptoms better or worse:

> Cecilia: I think the same, but I think environment is quite important. It's, like, you could, you could have been born with it *[autism]*, **but then the environment you were in could have made it worse or better**. (FGD6)

This argument suggests that participants distinguished between environmental factors that *cause autism* in the first place and those that may *impact its severity*. For instance, when they discussed the potential causal role of 'cold mothers' and 'bad parenting' on ASD (Table 6, s4.3.4.2), most participants found it difficult to believe that these could *cause* autism. Some mentioned that as

loving parents may help an autistic child cope with their symptoms, uncaring parents may worsen the child's traits, but this could not cause autism *per se*. An exception was FGD3, in which participants emphasized the way in which people are brought up (e.g. childhood neglect) as a risk factor for autism:

> Caroline: But then cold mothers probably would have to be there as well. Because that is bad parenting.
>
> Poppy: Yeah, it's been, like… um, um what's it called? Magnetic resonance… MRI scan of a neglected child's brain compared to a non-neglected child brain and that was, like, **a child's brain who hadn't been, like, loved by their mother basically and that was physically smaller and it was just horrible…**
>
> […]
>
> Alice: That could be like a stressor.
>
> Caroline: Yeah.
>
> Sophie: Or they're never socializing the kids.
>
> Alice: Yeah, like, neglect.
>
> Sophie: So when they do step out in public it would be a form of anxiety where they may become oversensitive of the surroundings, because it's 'oh my gosh, I have to talk to people and I don't know how'.
>
> Poppy: I don't, like, it depends… there is teaching of socializing and there's, like, actually socializing with the babies and… I think it's more not having taught how you say this to a person, **it's more like literally babies' brain is not developing because babies' brains I guess want love and people don't give it to them.**
>
> Interviewer: I see.
>
> Poppy: I think that's more my view rather than teaching them.
>
> Alice: Yeah, yeah, yeah… definitely.
>
> Caroline: I think your point was more… was like that, wasn't it? That you wouldn't know how to behave not because they haven't been taught, but because they haven't experienced it.
>
> Poppy: **I was thinking more like the physical, like the brain development**.

Poppy's comments in this quote introduce the second way in which participants reconciled their biological account of autism with environmental explanations, i.e. that environmental factors may impact children's brain development at the very early stages of life. This argument was often supported in relation to 'complications during pregnancy or birth' (Table 6, s4.3.4.2), which all

groups agreed do, or are likely to, cause the development of autism, as their friends' or relatives' experiences at birth would suggest.

Finally, some young people who were in the older age range and were more knowledgeable about genetics, reflected on how gene expression is influenced by environmental factors; one participant mentioned epigenetics:

> Virginia: **Environmental factors affect genes as well.**
>
> Interviewer: hmm, good point, yeah.
>
> Virginia: **'Cause, I mean, I'm doing a project on epigenetics so I…**
>
> Interviewer: Do you guys know what epigenetics is?
>
> Rose, Clara, Maureen: No. *[giggling]*
>
> Interviewer: Can you *[Virginia]* explain it in very simple words?
>
> Virginia: eh, ok it's quite hard to explain.
>
> Interviewer: Yeah, it's just…
>
> Virginia: I think I suppose, you can explain it as... how different genes are expressed in different cells and that can be affected by environmental factors making one gene more likely to be made into a protein so that [unclear]. (FGD2)

In some groups, increased knowledge about genetics or familiarity with autism seemed to be correlated with increased scepticism about the role of genetics in the development of autism. Indeed, there was a minority of groups (three out of 12) who proposed a less biological account of autism. In one of these, one participant had a brother with Asperger syndrome, and at the beginning of the discussion about ASD aetiology he argued that autism must be caused primarily by environmental factors because none of his other family members have the condition, and so they cannot have passed it onto his brother genetically:

> Jason: What I was going to say is genetics wise, I mean I'm not too sure. Neither of my parents have autism, but my brother does. I don't know how far back necessarily you can look at genetics until it becomes irrelevant to an extent. Like how many generations these go back. **But as far as I'm aware my brother is the only close one that I know with it. I don't know of any parents, grandparents or even great-grandparents who have had autism.** So I'd be tempted to move it towards the environment side more. (FGD10)

In a different FGD, Joanne made the opposite argument. Commenting that although she has a number of brothers, only one has autism, she reflected on the shared environment they all grew up in and concluded that autism must not be caused by the environment, but by genes:

> Joanne: Well, as my other brothers have been around **the same environment the whole time and they haven't really developed it** I'm thinking that genetics do play a part in it. But they can't just be the environment. (FGD8)

This suggests that in their dimensional and multifactorial understanding of autism aetiology, young people sometimes oscillated more towards the environment or the genetics end of the continuum. As explained, extensive evidence has shown that lay people construct their understanding of science and genetics on their pre-existing knowledge (the 'constructivist' rather than deficit model of lay expertise in genetics, see Kerr et al., 1998). Such knowledge is embedded in relevant aspects of people's social contexts, e.g. their family experiences as in the two quotes above, which may have primed Jason to focus on the environment and Joanne on genetic susceptibility. Moreover, despite their awareness of multiple risk factors, on some occasions participants seemed to 'strategically' select the causal factor that best served their intuitions or the argument they wanted to support. For instance, in the exchange in Box 7 (lines 20–28) Scarlett, who holds a strong biological understanding of ASD aetiology, quickly discards potential environmental factors that she mentions herself ('starvation of oxygen', line 25) to focus on genes and so reaffirm her position that 'it [autism]'s clearly only to do with genetics' (lines 38–39).

Although these argumentative strategies are common to the way in which people reason around heterogeneous and complicated phenomena (Condit, 2019), it is important to point them out, as they will help us interpret young people's discussions about interventions to prevent autism at different life stages. In the next chapter, I will show that there were instances in which young people were not always reflecting back on the dimensionality of ASD aetiology that they had constructed; on the contrary, like Scarlett in Box 7, they made arguments that seemed to embrace more 'comfortable' genetic deterministic views.

Comments revealing deterministic understanding of inherited patterns in autism were also more consistently expressed by groups with younger participants, which confirms findings from past studies on young people's understanding of inherited conditions (McGill et al., 2019) and will explain their attitudes towards interventions for the primary and secondary prevention of autism (Chapter 6). This is shown by the example below, where young people's reasons for arguing that autism is caused by multiple genes do not fit current understandings of dominant and recessive modes of inheritance, incomplete penetrance and variable expressivity:

> Luca: Multiple genes.
>
> Gabriel: I think it's a combination.
>
> David: It's a combination of multiple ones because **if there was just one there'd be a lot more people**, in theory.
>
> Matthew: I don't know.
>
> Gabriel: I think it's a special combination...
>
> David: A combination because people with autism, when they have children, **they don't always pass on that gene**. I think it's a combination.
>
> Luca: Yes, because otherwise **everyone who is autistic, their children would always be autistic as well**.
>
> David: Because if you just have that one autism gene **it would mean that one of your parents would definitely be autistic**. (FGD12)

To conclude, compared to the activity on ASD traits, most participants had less strong views about risk factors for autism. This is not surprising: given the increasingly recognized complexity of autism aetiology, which scientists themselves still struggle to grasp, some degree of speculation among lay people is to be expected (Qi et al., 2015). Within such speculation, young people sometimes expressed views that have been disproved, such as the link between bad parenting or cold mothers and ASD, which I addressed to ensure that participants did not leave the discussion with stigmatizing beliefs. Nonetheless, while negotiating ideas to reach a consensus, they ultimately endorsed a complex account of ASD aetiology, as caused by multiple genes that interact with the environment. In this way, most groups did not hold a genetic deterministic view of autism. From an educational point of view, this is where I had hoped my participants would get by the end of this

activity so that they could have nuanced conversations about the ethics of interventions to predict, prevent and treat autism.

5.5. Summary: Autism as a dimensional condition

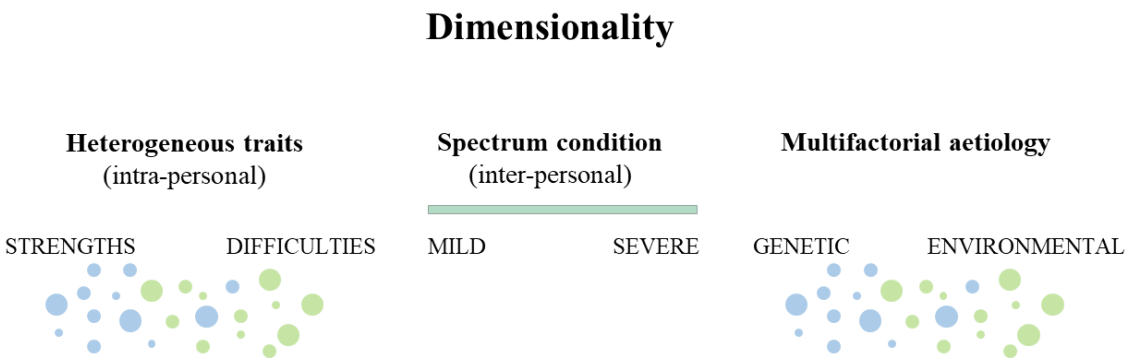


Figure 11: The threefold dimensionality of autism

The analysis of the data on participants' views of ASD traits and aetiology showed that young people co-constructed an account of autism that keeps together different components (strengths and weaknesses; severe and mild autism; genes and environment). Not only does this account match the various layers of complexity that characterize ASD (Chapter 3), it also indicates a threefold dimensional understanding of autism (Figure 11).

Some young people affirmed that everyone could be said to be somewhere on the autism spectrum, because everyone has some autistic traits. This suggests that they saw a continuum between autistics and neurotypicals, hence problematizing the concept of 'normality' and 'normal' behaviour. In the quote below, Jason conveys this concept through a metaphor, in which he compared autism to a jigsaw puzzle:

> Jason: Well, the way it *[autism]* was explained to me once was if you imagine, say, a 100-piece jigsaw puzzle, someone with autism is someone who has maybe sort of 30/40 pieces

> of that jigsaw puzzle anywhere from there upwards. Everyone may have… And imagine each piece to be like a separate symptom of it. **Everyone may have one or two pieces but in order to have autism it's a collective amount of those pieces.** That's how it was explained to me once. (FGD10)

That young people have a dimensional understanding of autism is important for two main reasons. Awareness of dimensionality is at odds with recent studies on lay knowledge and understanding of autism, in which participants hardly mentioned the autism spectrum and instead tried to provide a unified description of the condition (John et al., 2018). However, as I will discuss in Chapter 7, the educational activities I engaged young people in might have probed them to think of autism in a nuanced and dimensional way, even though to minimize any potential biases the activities on ASD traits and aetiology started with a brainstorming exercise.

More importantly, young people's dimensional conception is consistent with the recent shift in psychiatry to a recognition of the dimensionality of psychiatric conditions (Kong and Singh, 2018), including autism, as reflected in the changes introduced by the DSM-5. It also resonates with the plasticity that children with and without ADHD seem to attribute to their brain (Singh, 2013), despite worries that exposure to neuro-centred discourses may reduce their sense of agency towards a hegemonic and fixed brain. In the next section, I will discuss how the dimensionality which participants attributed to autism impacted on their discussion about the nature of ASD, particularly their uncertainty about whether autism should be seen as a disorder, disability, or difference.

For the moment it should be noted that the nuanced account young people developed while negotiating different beliefs in the group activities helped them grasp the dimensionality, and then discuss the uncertainty, of autism. This seems to be an important finding about the kind of settings that help genomic citizens in the making to develop sophisticated views of a certain phenomenon, and provides further evidence to the argument that with adequate support children can have a degree of capacities to contribute to ethics debates that resemble (or may even be better than) those of adults' (s2.4.1).

5.6. Disorder, disability, or difference? Uncertainty over the nature of autism

The question about the nature of autism was formulated to assess where young people would position themselves in the debate, discussed in the background chapters, about the different models of autism. In this thesis the term 'disorder' refers to the medical model of autism, the term 'disability' to the social model, and the term 'difference' to the neurodiversity model (s3.4). However, data analysis identified a discrepancy between these meanings and young people's understanding of the three terms. Participants often used the words 'disorder' and 'disability' interchangeably and in opposition to 'difference', and many young people had never heard of the concept of neurodiversity, as I will explain in Chapter 6. Thus, the discussion about the nature of autism often led young people to problematize the meaning of the three terms.

Section 5.6.1 below shows that overall young people expressed uncertainty about whether to conceptualize ASD as a disorder, disability, or difference; 'uncertainty' is therefore the category that emerged from participants' discussion over the nature of autism. Even though young people were asked a descriptive question (Would you say that someone who has all these traits *has* a disorder, a disability, or just a different personality?), in most cases their uncertainty meant that participants tested out different options about the way in which autism *should be* conceptualized. I argue that young people's uncertainty can be explained by their understanding of autism as a dimensional condition. In section 5.6.2, I show that, within their uncertainty, young people expressed some resistance against describing the condition as disorder or disability due to concerns about stigma attached to the two terms. This led some groups to strategically focus on the strengths that they associated with autism, to 'defend' autism as a difference. However, in stressing autistic advantageous traits, they abandoned the dimensionality of ASD and ended up promoting stereotypical and stigmatizing views about autism.

The following passage from FGD3 grasps young people's uncertainty about the nature of autism (Box 8). Uncertainty required the four participants (Poppy, Alice, Sophie and Caroline) to test out different opinions in a process in which stigma-related concerns were expressed, but a definitive or solid consensus was not ultimately reached.

Box 8: Young people's uncertainty about the nature of autism

1I: Do you think that people who have autism have a disorder or maybe a disability, or a different way of being?
[…]
4P: I think they need extra support, that's why we were annoyed because um… we thought, like, 'his school needs to know', like he's having problem making friends, obviously with the sensory… hmm, he would find that very stressful *[Poppy refers to a child she babysat]*, and… **but I think having the strength as well… whether it's like a full disability? It's just that they do need help, you know?**
9A: I just think that because society has told us that we are all kind of around that *[we all have a similar range of levels of ability in different tasks]* so anyone who is slightly different is seen as abnormal, so I think, like… **it's just a difference**, like they're just very []
12S: It depends…
13A: It depends on the severity, I think, as well.
14I: What do you mean?
15S: **It depends also on what you want to classify as a disorder**. Does it mean 'needing extra help'? because if that's the case we'll start calling dyslexia… wait, do you call dyslexia a disorder? I don't know, like…
18I: It's… again there are different opinions.
19S: If you're not very good at maths for example, it's not dyslexia, it's just your ability… would that be a disorder? Like, how far are you going to go?
21A: Yeah.
22I: hmm, ok.
23S: So yeah, that's one of the things which confuses me.
24I: Yeah.
25S: **And also the severity**, like some people don't really need all that much help and they can get into the adult world and be autonomous just fine and then there are some people who, they need to be accompanied throughout their lives because any small changes it's a complete difference. Um...
29I: So would you consider the second case as a disorder? And the first one as []
30**S: I don't know where the cut-off line is.**
31I: Ah ok, yeah.
32S: I don't think there is one.
33A: **That's why is so hard to []**
34P: Yeah, I think because it's a **spectrum** []
35A: Yeah.
36P: [unclear] A tiny little bit worse than that like []
37C: Because everyone's [unclear] it's hard to say well you have a disorder, but this person doesn't… like you don't []
39S: **Or you can say it's all a disorder but disorder shouldn't necessarily mean you need help.**

41C and A: Yeah.
42A: **It shouldn't be necessarily a bad thing.**
43S: Yeah, because you're going to give a definition for what a disorder is…
44C: I think it's just difference, but not… a disorder.
45A: Different, but not necessarily bad.
46C: Yeah.
47S: Difference, but not disorder.
48P: Yeah, I think obviously it's just when… like… when they, when their level of like… what's the word, what's the phrase? Their life um…
50I: Quality of life? No?
51P: Quality of life! Thank you. **When their quality of life is, like, so severely affected by autism** []
53A: **It's being unable to function, like, normally, or like, day-to-day life, yeah.**
54P: I think that's where, it's kind of the, it's horrible *[giggling]* and it's horrible for their parents as well, [unclear] really struggle, yeah so… it's not nice.
56I: Yeah.
57**A: But it's hard to define.**
58I: To define… yeah, yeah… but are you saying that in those cases then you would see that more as a...
60**P: hmm… I don't know… it doesn't mean like they're bad or autism is bad** []
61**A: I think, once it gets to a certain point, it should be classed as something because** []
62C: Yeah, because it's not []
63S: Because [unclear] **the help.**
64A: Yeah.
65P: I think they, it'd be like an identification.
66**A: Because otherwise they wouldn't get helped.**
67I: Yeah that's what I was going to ask. Do you think that, um, calling, maybe at least some cases as disorder has any kind of benefits?
69A: I think… I don't know.
70C: I don't think calling it a 'disorder' []
71A: No []
72C: It's helpful []
73A: But having a diagnosis helps get, helps them put mediums in place to ease life.
74C: Making the spectrum more like quantitative, rather than, like, 'oh you're all more severe than this'.
76**P: Maybe having the disorder, like, permeates like social stigma against** []
77S: That doesn't mean we should ban or stop using words, because**, you can always change people's perceptions of words.** It's not like, 'oh suddenly this is a bad word, ok let's not use that anymore'… like, no, just try and associate it with different things or accept…
80P: But I think that's hard for people, **I mean, because calling it disorder is put with such a wide variety of really bad things, if you get me? Like…**
82A: It'd be easier to change the name, like, the word.
83C: The word disorder is literally like…
84C and A: Dis-order
85C: They are not, I don't know how to explain it, **but it's not a nice word to use for it so, I don't know if it's the case of changing people's perceptions of it, I think it's changing the word for it.**

5.6.1. From dimensionality to uncertainty

As the interaction in Box 8 shows, uncertainty over the nature of autism was both implicit and explicit. In its implicit form, the category of uncertainty has a descriptive connotation, which is meant to capture the variety of responses that participants gave about the nature of autism. This means that within the same group different people held different views on whether autism is a disorder, a disability, or a difference; the same individual may have inconsistently described autism as a disorder, a disability, or a difference; or they may have changed their mind after listening to other group members. For instance, Alice initially seemed confident in her view that autism is a difference but is considered 'abnormal' because it causes behaviours that do not fit the range of traits that most people show. However, she quickly changed her mind when Sophie interrupted her to question her position ('it depends', line 12).

In its explicit form, uncertainty means that group discussions often culminated in participants' acknowledgement of the intricacy of the topic ('It's hard', 'I don't know', etc.), and of the reasons for their uncertainty, which encouraged groups to test out different options in which autism *should be* conceptualized. This is therefore the normative side of the category of uncertainty. In particular, participants used their discussion about the spectrum nature of autism and heterogeneity of ASD traits to explain their uncertainty, and so argue that there is no one-size-fits-all answer to the question about the nature of autism.

In the interaction in Box 8, uncertainty seems to derive primarily from the different degrees of severity that ASD presents ('it depends on the severity, I think, as well', line 13; 'and also the severity', line 25). This makes Sophie argue that it is impossible to identify the 'cut-off line' (line 30) where autism ceases to be a difference and becomes a disability or disorder. A similar argument is offered by Marta in a different focus group:

> Interviewer: Is that why you were saying that, earlier you were saying that it's difficult to kind of, like, classify autism?

> Marta: Yeah, **because it's so different**, like, there isn't – it's not like – **you could have somebody who you can barely notice it** and **somebody who is - you know, it really impacts them a lot** and they're, like, extremely different to the average person. (FGD9)

In the exchange in Box 8 (lines 4–8), Poppy introduces the link between heterogeneity and uncertainty when she questions whether ASD should be considered as 'a full disability' given that, in addition to difficulties that require support, autistics have strengths ('having the strength as well'). Thus, one participant suggested that because calling a person autistic is as broad as saying that they are female or male, autism should not be attributed a definition of either disorder, or disability, or difference ('it feels wrong', which again seems to be a value-laden, and so normative, judgement):

> Jason: **I suppose in a sense it's sort of saying someone has autism is similar to saying someone's a male or a female**. Like, there's so… You can have so many different types of people who are male, it's like you've got all sorts of different types of people, in a similar way there's also different types of people with autism, **so it feels wrong to put such a general, like, to put a term on such a general thing**. (FGD10)

Given that the heterogeneity of ASD traits and spectrum of severity mean that people have different experiences of autism, according to a few participants each autistic individual should decide how they want to define their autism depending on how the condition affects their quality of life:

> Evelyne: **It probably depends on how the individual wants to define it** and also how [unclear] because I feel like a disability is something which kind of negatively impacts you, like everyday life in some kind of way, so some people, they might find that being autistic is something they find like a very negative experience that they think they might be better off without, while other people I think would probably not like that terminology and think actually it's a kind of gift or something, I think. (FGD4)

This emphasis on self-determination, which as I will show emerged again in the discussion about treating autism (Chapter 6), might not only be because, given their young age, participants considered processes of experimenting, discovering and affirming one's own self as important. It may also be explained by young people's exposure to the recent public discourse and activism around identity politics based on gender, race, religion and so on (see Bragg et al., 2018), as Jason's

above comment could suggest. The application of this argument to autism in the context of my research is interesting because participants did not seem to be aware of similar forms of activism around ASD; indeed, as I will show in the next chapter, many participants had never heard of the concept of neurodiversity.

Given the above examples, I argue that the two subcategories of 'heterogeneity' and 'spectrum' identified in the first part of this chapter, and so young people's conceptualization of autism on a dimensional continuum from strengths to difficulties and from mild to severe forms, explain why no strong belief was expressed about whether autism should be categorized as a disorder, a disability, or a difference, but rather a variety of opinions were proposed. As Box 8 shows (lines 29, 58–59), during data collection I reacted to young people's uncertainty by asking whether they considered severe forms of autism as a disorder. A subset of groups were asked to respond to the profile of Sam (Appendix M), a 16-year-old autistic boy who is non-verbal, has learning difficulties, requires constant support and supervision, and self-harms or uses violent behaviour as a coping mechanism when he gets agitated. Because I had approached the study of autism in a dichotomous way, by asking about Sam's and other severe cases of autism I expected that participants would take a clear-cut position about the nature of ASD.

When challenged in this way, some participants did argue that the more severe the autism is, the more it should be regarded as a disorder or disability. As Daniel from FGD6 put it: 'When it's more severe and it takes over, like, more of their life, it's definitely more of a disability.' However, this was not always the case, since some groups resisted attributing those terms to autism. As I show in the next section, this was because of concerns about stigma attached to the two terms, which confirms that the FGDs gradually shifted from a focus on participants' factual knowledge to a normative discussion that allowed young people's moral concerns to emerge.

5.6.2. Adopting dichotomous thinking: From uncertainty to stereotypes about ASD

As mentioned above, in the discussion about the nature of autism young people problematized the meaning of the terms difference, disorder, and disability. Table 8 presents the definitions that participants co-constructed.

Table 8: Participants' definitions of difference, disorder, and disability in relation to autism	
Difference	Different way of thinking, which may be perceived as wrong and difficult to understand because it is unusual and does not fit societal expectations to what 'normal' is.
Disorder	A disorder affects a person's quality of life, causes distress, disfunction and an abnormal development, it makes a lot of things harder, it is very limiting and does not have any positives attached to it.
Disability	A disability causes people to be unable to do things, including to provide for themselves, it causes them to being incapable, and negatively impacts people's everyday life. A *mental* disability is a disability that is linked to the person's brain structure and development, cannot be cured and makes people unable to understand the impact of one's actions on other people.

These definitions suggest that young people attached a strong negative connotation to the terms 'disorder' and 'disability', which as mentioned were used as synonyms. Such understanding may be close to the everyday use of the two terms (Kahane and Savulescu, 2009), but the more extremely negative evaluation that participants in this study attributed to the two terms reveals stigmatizing and stereotypical assumptions about disorders and disabilities ('causes distress, disfunction and an abnormal development', 'being unable to do things', 'being incapable', etc.).

This may explain why, upon my request to clarify whether they thought that severe autism should be considered as a disorder or disability, some young people resisted attributing those terms to ASD due to concerns that this could increase stigma towards autistics. In some cases they reiterated their uncertainty regarding the nature of autism, like Poppy in Box 8, line 60 ('I don't know… it doesn't mean like they're bad or autism is bad'). Some participants did so to suggest that all forms of autism are valuable *per se*, and so the severity of autism is irrelevant to discussions about the nature of autism. This attitude towards autism is well captured by Gracie's comment below and, as I will

explain in Chapter 6 (s6.5), was used by a minority of participants to argue that reproductive choices about testing and abortion should not be about autism in the first place:

> Interviewer: Do you know anyone with severe forms of autism?
>
> Gracie: I don't know, I've never asked how bad their autism is. I've just accepted they've got autism. (FGD7)

Thus, although in my questions I often presented young people with binary options (in this case mild vs severe, difference vs disorder/disability), in their responses young people proposed alternatives that did not fit the dichotomies that I presented them with. I will further comment on this point in the concluding remarks.

It could also be the case that young people developed a negative account of 'disorder' and 'disability' because they wanted to distance themselves from stigmatizing views of autism. In these cases, expressing stereotypical views about the two terms served the purpose of constructing a positive narrative around autism, because people with autism do not fit the negative definitions presented in Table 8. This is illustrated by the example below in which to argue that there is nothing 'wrong' with autism, Greta seems to imply that disorders and disabilities are not 'normal':

> Greta: It's hard, it's kind of like a yes and a no kind of thing because, yes, in terms of medical things they do have a disorder or disability kind of thing. But as you said earlier about labels, it can make it seem like there's **something wrong** with them when there isn't really. They're just a **normal** person. They just think differently. (FGD8)

When young people wanted to 'defend' autism and present it as a difference, they tended to emphasize autistic strengths and special talents:

> Cecilia: I think that, like, everyone has their, like, things that they're good at and not so good at. It just shows, like, **how they're maybe a lot better than us at some things and then they might really struggle in other aspects**. And I think that just means that they're **different**, not that they have a disability, that they are just different to all of us. (FGD6)

This seems to be an example of how young people strategically cherry-picked different bits of their knowledge of autism, in this case autistic strengths, to their own argumentative advantage, i.e. to

argue that autism is just a different part of the variety of human traits. In the quote below, Ludovica seems to suggest that autistic traits should be celebrated because autistics could be people like the mathematician Alan Turing:

> Ludovica: I think more needs to be kind of **celebrated** about these **strengths** of autism, like the film The Imitation Game. **Alan Turing** was autistic and if it wasn't for him, like, the world would be completely different because he helped so much and it's people, kind of… there is such a set stigma around autism as some of the negatives and people just think of that rather than looking at the…
>
> Interviewer: The positives.
>
> Ludovica: Yeah, so I think there's more to celebrate. (FGD1)

In so doing, participants ended up constructing a narrative of autism that is equally stereotypical to their narrative about disorders and disabilities, albeit in a different way. They seemed to place the value of a person with autism in their special talents, or they granted autism the status of 'difference' only because people with autism may be 'a lot better than us in other things', as Cecilia says in the above quote. Some authors have identified the same reasoning in media representations of autism, in which autistic people' lives are shown as valuable because of autistics' savant skills (see Murray, 2008, cited in Draaisma, 2009).

However, in this study resistance against attributing the definitions of disorder and disability to autism did not equal refusal. When they problematized the meanings of these two words participants proposed three arguments to suggest when autism could or should be called disorder or disability. The first refers to the practical advantages that an ASD diagnosis could provide autistic people with. Defining autism as a disorder or disability could help people get the support they need, which would not happen if autism was only perceived as a difference, and may help society be more tolerant towards autistics and more respectful of their needs:

> Ludovica: I think sometimes it is helpful to say that it is a disorder or a disability because then **if they need help in schools or in their life, there is going to be more support** for them, whereas if you label it just as personality traits then people are **less forgiving** when there is a struggle with it. (FGD1)

The other two arguments are suggestions about alternative meanings that the terms could be attributed. Autism could be defined as a disorder because it does cause some difficulties that cannot and should not be neglected. However, 'disorder' does not necessarily have to be attributed a negative evaluation or, as Alice says in Box 8 (line 42), 'it shouldn't be necessarily a bad thing'. This argument recalls attempts to transcend the polarization between different models of autism (s3.4.3) and accept autistic deficits as differences that may be disabling at times, but are also valuable parts of human variety around which a positive identity can be built (Kapp et al., 2013). Finally, one could call autism a disorder or disability in the sense that autistic behaviours and traits show a way of functioning that is different from that of most people:

> Eloise: I guess it's like you said on the spectrum because there are some aspects of it which do disable them in a certain context from functioning adequately. It's, like, in psychology we did the **deviation from social norms** and the different definitions of that sort of thing. So it **doesn't mean they're necessarily disabled**, it just means that **they're deviating from social norms** and stuff like that. (FGD10)

This definition aligns with the 'biostatistical theory' or 'naturalistic account' (Boorse, 1977), which understands normality as a biological, statistical and value-free concept, and considers disorders and disabilities as a statistical deviance from human typical development or functioning. In this sense, some young people argued that neurotypicals may perceive autism in a negative way because autistics show behaviours they are not used to.

In summary, section 5.6 showed that participants expressed a variety of opinions about whether autism should be seen as a disorder, a disability, or a difference, which mirrors the complex views that exist in the public debate around the nature of autism (Chapter 3). Data analysis showed that this uncertainty is due to the heterogeneity of autistic traits and the spectrum of ASD severities. Thus, as Figure 7 illustrated at the outset of this chapter, the dimensional understanding that participants have of autism explains their uncertainty over the nature of the condition.

What does not feature from Figure 7 in young people's discussions about the nature of autism is the third component of the dimensionality that young people associated with autism, i.e. the idea that autism is caused by factors across a continuum from genetics to the environment. Indeed, in this activity considerations about autism aetiology were not made. Thus, despite groups' preference for a biological understanding of autism they did not straightforwardly adopt a medical model that regards autism as a disorder. There was only one instance in which one participant suggested this link, although he clarified that he disagreed with it: 'If it's genetics, it kind of is a disorder but you wouldn't think of it like that.' (Ted, FGD12).

This may be because strong biological accounts were mitigated by the acknowledgement of environmental factors, which in turn suggests that, although not explicitly, perhaps implicitly the dimensionality of ASD aetiology contributed to create a complex account of autism that fed into young people's uncertainty. However, it could also be that young people considered details about the biological underpinnings of ASD as irrelevant to the way in which autism should be conceptualized. In other words, the increasing application of the 'genomic gaze' (Singh, 2016b) to the study of autism in the last few decades, to which I referred on several occasions during the focus groups, seems to have had little impact on the way in which young people from the general population think of autism. Not only does this question the argument that genetic citizens reduce differences between people to their genes (Lippman, 1991), it also casts doubts on the assumption that exposure to genetic words, concepts and explanations make lay people genetic essentialists – if anything, they might be 'strategic' essentialists (Condit, 2019) or determinists, as seen above.

It was only when one side of the ASD dimensionality (i.e. strengths) was accentuated that a more polarized view of autism was expressed: a small minority of participants more strongly supported an account of autism as difference, and in so doing some made comments that revealed stereotypical assumptions about autistic talents and about where the value of autistic lives should be placed.

Notably, no group challenged uncertainty by leaning towards ‘disorder’ or ‘disability’, probably because of the highly negative connotation the young people attributed to these terms.

5.7. Concluding remarks

This chapter sought to analyse young people’s views of autism, i.e. their understanding of autism and autism aetiology, and their conceptualization of the nature of ASD. The above sections have shown that when conducting data collection I was moved by a binary way of thinking, which is reflected in the dichotomous questions I asked, while in their responses young people often moved beyond the dichotomies. While analysing the data I realized that the binary thinking that I was trying to apply does not fit with the account of autism that young people co-constructed. Indeed, two main categories, ‘dimensionality’ and ‘uncertainty’, emerged. In this chapter, I showed that young people’s conceptualization of autism in a dimensional rather than dichotomous way explains why they did not hold strong views on whether autism should be considered as either a disorder, or a disability, or a difference (Figure 7).

Young people’s endorsement of dimensionality and uncertainty in this first investigation of the ethical issues that genomic citizens ‘in the making’ express at the intersection between genomics and mental health is important, because it is consistent with the various layers of complexity that this field entails, as discussed in Chapter 2. However, the philosophical analysis of participants’ contribution demonstrated that at times young people abandoned their dimensional account of autism and uncertainty about the ASD nature and became ‘strategic dichotomists’ – after Condit’s ‘strategic essentialists’ (2019), i.e. they adopted more ‘comfortable’ dichotomous and clear-cut views about ASD traits and causes. In so doing, they promoted myths and so stereotypical and stigmatizing views about autistic special talents, which could trivialize the suffering that autism causes to some people and their families; or genetic deterministic views about ASD aetiology, which may lead to misinformed choices about autism genomics.

At the beginning of this chapter the investigation of young people's views of autism was deemed necessary to interpret their attitudes towards the development of treatments for autism, genetic tests to prevent autism prenatally and genetic tests to perform early interventions for autism. In the next chapter, I will critically present such attitudes, and I will show that in the ethics discussions about the three interventions, some 'strategic dichotomies' were deployed again, hence promoting deterministic or stigmatizing views. This confirms the importance of making genomic citizens comfortable with embracing dimensionality and uncertainty.

Chapter 6. Young people's attitudes towards using ASD genomic information

6.1. Introduction

Chapter 5 presented young people's views of what autism is and is caused by, and how the nature of autism should be conceptualized. I demonstrated that young people developed a dimensional account, which enabled them to accommodate their uncertainty over whether autism should be seen as a disorder, a disability, or a difference. In this chapter, I will show how young people's views of autism help to inform their attitudes towards three interventions for the primary and secondary prevention of autism, i.e. the development of pharmacological treatments for autism; prenatal diagnosis (PND) and preimplantation genetic diagnosis (PGD) to prevent autism before birth; and genetic tests performed on young children to assess their susceptibility to autism for early intervention purposes. In particular, this chapter seeks to tease out the arguments underpinning young people's attitudes towards these interventions, by addressing three sets of questions:

1. Should we welcome genomic research to develop pharmacological treatments for autism? Why?
2. Should parents be allowed to use genomic information about their embryos' and foetuses' predisposition to autism to make reproductive choices? Why?
3. Should we test babies for their genetic predisposition to ASD for early intervention purposes? Should we enrol high-risk children in early intervention programmes? Why?

6.2. Overview of the present chapter

As explained in Chapter 4, a discussion about the concept of neurodiversity and the neurodiversity model of autism served as a link between activities that focused on young people's factual

knowledge of autism to those that investigated their moral attitudes towards preventing autism at different life stages. Section 6.3 will explain that, although young people believed that society should do more to accommodate autistic traits, they pointed out that some ASD traits are intrinsic difficulties, which cannot be said to be caused by societal barriers, and they discussed a range of reasons for why society may find it more difficult to accommodate psychiatric conditions compared to non-psychiatric conditions. Therefore, participants expressed a presumption in favour of allowing people to make their own decisions about the interventions for the primary and secondary prevention of autism, which requires that these are made available.

In sections 6.4 to 6.6, I will show that despite such presumption, young people's description of autism as characterized by heterogeneous traits across a spectrum, and so their uncertainty about its nature, as well as their multifactorial account of ASD aetiology, motivated a range of concerns towards using genomic information to prevent autism at different life stages, some of which focused on the availability of the interventions and others on their implementation. Across the different activities, similar categories were identified, revolving around the potential for imposing 'coercive measures'; the risk of encouraging 'stigmatizing' views of autism; the risk of reducing 'diversity' in society; the impact that the interventions may have on 'identity', both in terms of normalizing autism and of hindering children's opportunity for determining who they want to be; and the 'limited clinical validity and utility' of the interventions. These concerns motivated a minority of groups to interrogate the meaning of 'being a good parent'.

This chapter will anticipate that young people's arguments did not fit the traditional notion of biological citizenship, within which the responsible citizen is expected to draw on genomic knowledge to treat and prevent health conditions so to maximize health and minimize the risk of illness. Rather, young people's narrative suggests that the application of genomic science to mental health necessitates a shift to an expanded notion of biological citizenship, the features of which will be discussed in the next chapter.

6.3. Young people's attitudes towards the neurodiversity argument

To introduce the topic of neurodiversity, I asked participants whether they thought that autistic people should change themselves to overcome the difficulties associated with autism to function well in society, or whether society should put services in place to accommodate autistic needs; in this sense, the neurodiversity model was presented as close to the social model of autism (s3.4.1). Most groups (nine out of 12) were also explicitly asked whether they had ever heard of the concept of neurodiversity. This was not the case for many participants; however, young people showed that they could guess the meaning of 'neurodiversity' and reflect on whether they agreed with the model of autism proposed by the neurodiversity movement, which in this chapter I will call the 'neurodiversity argument'.

All participants agreed that society should do more to accommodate autistic traits. This was not only because they thought that some societal changes are simple. Most importantly, young people grounded their argument on their belief that autism should not be necessarily seen as a disorder or disability, but also as a valuable difference ('uncertainty', s5.6), and that autistics have a right to equal respect like everyone else. As Ruby from FGD11 put it: 'If people are different, we want them to be treated with respect. And to be understood for who they are and not to change that'.

Moreover, participants argued that a diverse society tends to be more tolerant and to function better, as it benefits from people's different voices and perspectives. To support this claim, some participants resumed their stereotypical assumptions about autism, when they affirmed that autistic people in particular make a valuable contribution to society thanks to their special talents:

> Leo: […] And it would be much easier to just change the perceptions and opinions of those around them *[autistic people]* to show them that they're normal in the way that they are. They just were born with slightly different traits that led to them, to act the way they did.
>
> Ruby: I don't think they're necessarily normal. **I think it's amazing, there are great artists and literature people who have autism and have changed how people view certain things.** That is amazing! I don't think they should ever be seen as something [unclear], someone says 'you have to change how you are', because everyone is different

> in a certain way. And I think that's the best thing about it. I genuinely understand where you have to… that some people need certain needs more than others. But at the same time, **I don't think you should ever really have to change someone like that, because people change the world.**
>
> Leo: Yes, that's what I'm trying to say.
>
> Scarlett: **Diversity is good.**
>
> Ruby: **It's amazing.**
>
> Scarlett: Without diversity, everyone would just be the same.
>
> Leo: It would be quite boring.
>
> Scarlett: Yes, it would be boring but also, I think **the more different sorts of people there are, certain, the more accepting that everyone is of each other.** Everyone's different, otherwise if we were all the same then we'd actually never understand people who do something with disabilities. (FGD11)

Notwithstanding their overall support, participants expressed reservations about the neurodiversity model of autism; as Alice from FGD3 argued: 'I think the whole concept is… is it a good idea to raise, or just to say that not everyone is the same, there is diversity, but it's hard to know what to do with that fact.' From the analysis two main limitations emerged. First, some participants remarked that society is not necessarily responsible for causing some autistic difficulties, such as hypersensitivity to stimuli, which are instead 'intrinsic limitations':

> Maureen: Yeah, no, I agree when it says that, hmm, 'the negative consequences of having an impairment often come from societal structures rather than the condition itself' *[see the leaflet on neurodiversity, Appendix I]* […] Whereas 'some autistic persons may react badly to lights or sounds that are common in the social world'. **I think it's not really society that's causing them to react badly though,** because it's like not anyone doing that, it's just light and sound so I don't think it's societal structures that cause them to feel left out. But if they react badly and then society says 'well then you are not normal' then it's society's fault, it is not the light and sound. (FGD2)

This argument aligns with the criticism that is often raised against strong social models of disability, particularly against their assumption that disabled people's social disadvantage is solely caused by societal barriers. As Tom Shakespeare has argued: 'In most cases, disabled people are experiencing both the intrinsic limitation of impairment, and the externally imposed social discrimination' (2014, p. 30).

The second limitation of the neurodiversity argument emerged when participants were encouraged to make comparisons between non-psychiatric conditions (e.g. hearing loss or blindness) and conditions that affect people's behaviour. When prompted in this way, they tended to argue that it is more difficult for society to accommodate traits in the case of autism or other psychiatric conditions for a variety of reasons. First, young people believed that the heterogeneity and spectrum nature of autism make responding to the needs of autistic people less straightforward. At times accommodating everyone's needs and removing all obstacles may not only be difficult, but rather impractical. Marta from FGD9 provided the example of her cousin for whom finding the right educational system has been challenging because 'she's like, yeah, she's not severe enough to go to, like, a special school but she's not neurotypical enough to go to a normal school'.

This confirms that participants understood autism as a dimensional condition that manifests on a continuum of traits and severities. It also suggests that intuitively young people drew a conclusion that scholars have objected to the strong version of the social model, particularly its ideal of a barrier-free world (s2.2.2.1). Shakespeare (2014) gave precisely the example of autism to illustrate the impracticality of adapting the mainstream world to accommodate, alongside other people, the needs of someone whose main difficulties reside in social interactions. However, he extended his argument to any disability characterized by heterogeneous traits on a spectrum, while the study participants considered these features as problematic only in the case of psychiatric conditions. This might be because young people had a positive, and arguably too optimistic, perception of how physical disabilities are treated in society, as this example from FGD11 suggests:

> Interviewer: You keep saying mental versus physical. Do you think it's different with physical, let's say, disabilities although we haven't agreed []
>
> Scarlett: I think so.
>
> Leo: **If, say, your legs didn't work there are many things that people can do to make sure that that has very little effect, overall, on your life**. Obviously, you wouldn't be able to walk and that is horrible. But it doesn't mean you can't go outside, it doesn't mean you can't go to a regular classroom. It just means, say, it is horrible that you can't walk. **You can still function and have friends and be part of society** even though you are born with, or say have due to an accident, this physical disability. **But with a mental disability,**

> **it's a lot more complicated. And it's a lot harder to treat especially because everyone is different.** (FGD11)

Regardless of the validity of the distinction that young people drew between psychiatric and non-psychiatric conditions, with which disability authors like Shakespeare (2014) would probably disagree, participants' pessimistic views of the way in which psychiatric conditions are treated in society was expressed on multiple occasions throughout the FGDs, and seemed to underlie the discussion of the other limitations that they identified in the neurodiversity argument. This includes the 'invisibility of autism' and other mental conditions, which makes it more difficult for society to accommodate the needs of people who suffer from them:

> Amelia: It's like… hmm… it's like how there are, like… say, with depression and **someone with a broken leg you can clearly see**… someone who has a broken leg, so they try to accommodate them so, like, there are lifts for people in the wheelchair and stuff, when someone has a bad leg in my class everyone has to go downstairs and not upstairs. **But then when someone has autism you can't see it when you immediately look at them so then it's hard to… like, include them kind of like make special services all the time, I guess**. (FGD4)

In addition, because autism affects people's personality, behaviours, identity, i.e. traits that are associated with 'personhood' (Silverman, 2008), according to the young people neurotypicals may struggle to relate to or interact with people with autism, and so to respond to their needs. This is particularly complicated because autism is a congenital and lifelong condition, given that it has to do with people's biology. Therefore, as Leo argues below, autism is a 'constant difference' rather than a temporary impediment in social interactions:

> Leo: If, say, you were in a wheelchair, besides the fact that you're in a wheelchair you're completely normal. And 99% of normal people can just say that they're the exact same as me. Their legs don't work or this is the physical problem with them. But with autism, it's a **constant difference**. **Every time you interact with them** you'd have to, not necessarily walk on ice, but **you'd have to act differently** to make sure that they're on the same page. Whereas with a physical disability it's the exact same person, they're just in a different state. And I think that'd be a lot easier for people to cope with. You'd have a lot better of a time with, say, a physical disability than, say, a mental one like autism. Not that either are desirable. (FGD11)

The fourth reason why young people thought that society may be less willing to meet autistics' needs was 'societal stigma', which according to participants is stronger in psychiatric conditions than non-psychiatric conditions. As the following passage from FGD4 shows, groups identified various ways in which psychiatric stigma manifests, which echo some of the arguments discussed in the literature (s2.3.2). Society tends to associate mental (ill) health with dangerousness ('people are scared of it potentially', 'crazy' and 'dangerous') and so, as Evelyne argues, desires distance from people with mental health issues. Moreover, lay people tend to blame individuals with psychiatric conditions as responsible for their behaviour, to which, in line with participants' biological account of autism, Jasmine counterargues that autism is instead 'part of their brain':

> Evelyne: People seem far more willing to accommodate physical illness or disability…
>
> Amelia: Yeah.
>
> Evelyne: …than mental ones, I think.
>
> Interviewer: And why do you think that's the case?
>
> Amelia: Because you can visually see it, maybe? You can see that something is wrong with them.
>
> Evelyne: I don't know **people are scared of it potentially,** like, I don't know…
>
> Interviewer: Scared of?
>
> Evelyne: People who are different.
>
> Interviewer: Yeah, but you mean…
>
> Evelyne: A person with a broken leg; anyone could have a broken leg, but people are kind of… well, obviously, anyone can have mental illness as well but they kind of, it is something that **they kind of want to keep a distance from** and not really that… I don't know.
>
> Interviewer: Yeah, go ahead.
>
> Jasmine: I think that it's **because it's a mental thing then a lot of people think that you can change it,** so they're, like, 'oh, it's just a state of mind that they are in' or… whereas obviously a broken leg you can clearly see that it will take this amount of time to fix. Whereas I think... so they think **'oh, autistic people should just get on with it and sort themselves out'** whereas it's not really something that they can… like, **it's part of their brain**.
>
> Interviewer: […] And do you have many… maybe a hypothesis on why people are scared of mental health issues more than physical conditions?
>
> […]
>
> Jasmine: I think because **there is such a stigma** and there is such… people really associate, like, any mental health disorder to '**oh, they are crazy', 'they'll be dangerous,**

> and I think that is a big reason why people are really scared of it because they are, like, 'oh, that person shouldn't be allowed out', 'they shouldn't be allowed in my movie theatre', because you know '**they are more dangerous than we are'** but that's not the case; **they are just not educated about it**.
>
> Evelyne: It seems like if something does happen, like **if one person, I don't know, stabs someone then the media immediately jumps on 'oh look, they have it',** but actually if you look at, probably, statistics the majority of people do not have any kind of problem and it *[crime or violent behaviour]* is probably not linked to it *[psychiatric condition]* anyway.

Finally, as Jasmine mentions in her last comment above, society may be less equipped to respond to autistic people's needs compared to those of people with physical disabilities due to the lack of education and knowledge about ASD. Thus, in other FGDs, participants suggested that improved understanding of autism could help people be more empathetic towards autistics.

To recap, young people agreed with the neurodiversity argument to the extent that they believed that society should do more to accommodate autistic people's needs. However, by arguing that some difficulties are intrinsic in autistic people they challenged the assumption of the most radical versions of the neurodiversity movement, according to which society is necessarily responsible for autistics' social disadvantage; and they observed that there may be little society can do to accommodate some autistic difficulties. Moreover, participants compared psychiatric and non-psychiatric conditions to identify various factors due to which society may be less willing or equipped to respond to autistics' needs. These include 'ASD heterogeneity and spectrum', 'invisibility', 'affecting personhood', 'psychiatric stigma', and 'lack of education and empathy'. Thus, despite their positive views of autism, young people expressed an initial presumption in favour of welcoming the interventions for the primary and secondary prevention of ASD. The next sections provide a detailed presentation of young people's attitudes towards these interventions.

6.4. Young people's attitudes towards treating autism

When discussing the thought experiment about the hypothetical pill for autism (see the dialogue in Box 9), young people argued in favour of welcoming genomic research to develop pharmacological

treatments for ASD and supported their views on the basis of considerations about autonomy and beneficence. However, they discussed several concerns that emerge from either the mere availability of a 'cure' or the decision to take the hypothetical pill.

Box 9: Thought experiment about the hypothetical pill for autism
Naoki: Hey Sue, have you heard? Scientists claim they have developed a pill to cure autism. Here it is: we could actually try it and see if it works. Sue: Wow, this is interesting… Naoki: I don't know… I used to think it'd be the best thing if I could just live my life like a normal person, but now I am in doubt whether I should take the pill. Sue: Why did you change your mind? Naoki: Well, autism is such an inseparable part of our identity. Wouldn't eliminating it (if this is even possible) mean eliminating who we are? Sue: Well, the possibility that I could be very autistic for the rest of my life always upsets me, so I would love to hear more about this pill. Naoki: Really? Sue: Yeah, you see, people with 'high-functioning' autism are different, but basically they are just a variation of the norm. Some even have exceptional abilities that should be celebrated and encouraged. But people with severe autism, like me, are just trying to get through the day without hurting, tapping, flailing, biting, screaming, etc. It is a constant struggle. For instance, when I have to mask autism in class it takes a tremendous amount of effort. So I think that a cure for autism, would be wonderful. Naoki: But isn't it a struggle for everyone? Every human being, with or without disabilities, needs to strive to do their best; and for us having autism is normal, so we cannot know for sure what 'normal' is even like… I think I might well choose to stay as I am.

Throughout the next pages, I will refer back to the following passage from FGD9, in which Simon, Marta and Adele weigh arguments in favour and against the use of genetic knowledge to develop a hypothetical pill for autism.

Box 10: Young people weigh arguments in favour and against treating autism

1S: Um, yeah, hmm, I think it's quite a difficult one *[Simon refers to the dialogue about the hypothetical pill for autism]*, because with this example, like where she's obviously trying to be, um, like, not hurt other people, and they're obviously understanding that they're hurting people, um, in this way, obviously **that would be sort of good if they could control it**, but then the – yeah, I don't know.
6M: **I think it should be a personal decision**, like, for the person. And **I don't think it should be something that parents can give to their children necessarily**, because I'm not sure, that's difficult, you know, **that's age of consent**. Because if they're medicated up until the age of consent, then how do they know whether they want to continue or go without it? Like, they would have just known it from an early age, so it's a bit difficult in that regard. But if you are considered to be able to make that decision by yourself, then I think you should be allowed to.
12I: And why do you think it's important that it's the person's decision? Because, if you think about it, parents give medications for many different things.
14S: Hmm.
15I: Like, if it wasn't the case of autism but about cancer, say, or heart problems.
16M: Because **I think ultimately it's to do with how it affects the individual**. Like, obviously it affects other people too, and you do have to take that into account, but it's the individual's, you know, disorder or however you want to classify it and, you know**, they should be able to choose what they think is best for them** and what will help them. And, yeah, **if you start forcing it on people** then it's not, you know, well, **it makes them feel horrible**.
21A: I mean, like, inevitably their life is theirs.
22M: Yeah, exactly.
23A: And beyond when your parents, obviously they're not going to be around forever, so therefore how, almost like how do they have the right to determine the path of their life. Because at the time it was their choice to decide whether the tablet was necessary, but then you could argue, **are the parents doing it for themselves** so, 'It's easier to look after my child because I've given them this pill', **or are they giving them the tablet to then say, 'It's easier for my child'?** So it's kind of, like, **who are you doing it for?**
29I: Hmm, yeah.
30S: Yeah, again, I don't know how – because if you're literally taking every aspect of this autism, um, away from the person, it's sort of like, I don't know, taking – it's sort of []
32M: **Changing who they are.**
33S: Yeah.
34M: A little bit, hmm.
35I: Say it again? Sorry.
36M: **Changing who they are, changing them a little bit.**
37I: Yeah, yeah. And what – no, go ahead.
38S: Yeah, it's just, so obviously with high-functioning people, as I was saying, it's like they can still go around and live relatively normal lives, just with differences. But when you have autism and they're hurting themselves or others around them, it's just, like, very hard, because you don't want that to be – **no one wishes for a child or someone to grow up, like, with something that's going to massively affect them** and their quality of life. **But just, I think, just taking it away and then suddenly being what is perceived as a normal person is such a massive change**, that I don't know if it's, like – I don't know how to describe it.
[…]
46M: I think **it also depends on what exactly this pill does and to what extent**.
47S: Yeah.
48A: Yeah.

49M: Because if it just sort of – because if it completely sort of like, like you were saying, sedates them and. Because is this routine helping them, like, focus and maybe stay calm and not get scared or stressed out about things and completely changing who they are and how they act?
52A: Yeah.
53M: You know, **it's like managing it or getting rid of it, like, which, what does it do?** And to what extent, how much does it change them?
55A: Yeah.
56M: So, yeah, I don't know. It depends.
57I: Yeah.
58M: Yeah.
59I: So would you be more comfortable with a pill that only addresses or targets some difficulties?
61M: Yeah, I think so. Because you don't want to get rid of all of it, because – yeah.
62I: But again, all these examples that you are giving make me think, like, **do you think just the availability of a cure or a pill might affect or change the way that society looks at autism or people with autism?**
65S: Exactly yeah. It's that sort of thing, it's, 'Oh, you can cure yourself', when you're not necessarily, like, wanting to change *[laughs]* yourself. It's like pressure.
67A: **Pressure that you'll then be given.**
68S: It's like…
69M: Yeah, it's, like, '**Oh, this is [something bad] so it needs to be fixed'.**
70A: Yeah.
71S: **So, 'Take this', rather than this is a thing that people have and people should sort of just work around** and sort of, like, just work with everything, rather than sort of, 'This is something you can have to stop this'.
74M: Yeah, I think perhaps it's to do with being, like, marketed as an autism drug. It could be something that doctors or whoever, you know, healthcare professionals just say, 'This can help you deal with any problems you might face, and it's not to cure your autism, it's just to help you manage some of the difficulties that come with it'. **So it's not seen as, like, a cure or a preventative measure or anything, it's just like a helping hand**, you know, another way to support them, if you know what I mean.
80A: Hmm, hmm.
81S: Yeah, yeah, so **I agree with things that help them medically, like, for instance, help with anxiety or something like that** []
83M: Hmm, exactly, that kind of thing.
84S: Will inevitably reduce sort of, potentially, like, breakdowns or whatever, meltdowns. Um, and I agree with stuff to help them with gastrointestinal stuff and things like that, **that should definitely be used**. And something, if there was a pill to stop them from, I don't know, being, in the severe cases, completely cut off or, like, making it easier for them to talk to people or, if they are violent, then if you can – like, towards themselves – that they can stop something, stop them hitting themselves or scratching.
90I: Or, like, head banging, yeah.
91S: Exactly. Then that would be definitely something. **But I don't think something to go, 'You can cure autism and then you're going to be someone just, like, different', is right.** I don't know.

6.4.1. Treatments could promote autonomy and benefit autistic people

In most groups, participants' initial reaction to the dialogue about the hypothetical pill for autism was to support the autistic person's individual and autonomous choice in the context of decisions around treating autistic traits. For instance, in the passage above Marta argues that 'it should be a personal decision' (Box 10, line 6). This means that participants' attitudes did not match those of the most radical neurodiversity activists, who eschew the 'cure-oriented agenda' of the medical model (s3.4); rather, for young people there was no strong enough reason to shun genomic research for the development of pharmacological treatments for autism.

Further, as Simon repeatedly affirms in Box 10 (lines 1–5, 81–82, 84–89), the development of treatments for autism may also provide autistics with medical and psychological benefits, for instance in the form of improved quality of life. This would be particularly the case if treatments only targeted severe difficulties, by which participants meant comorbid medical and mental issues such as 'anxiety', 'breakdowns', and 'gastrointestinal stuff'. In this sense, the young people seemed to appeal to the principle of beneficence. This discussion corroborates Shakespeare's argument (2014) that (disabled) people's attitudes towards medical cures depend on the *type* of medication at stake, which could aim at complete elimination of impairment; better life expectancy; improved functioning and quality of life; or normalization and cosmetic improvements. Although the study participants seemed to use the two terms interchangeably, their attitudes and arguments suggest that they saw a normative difference between 'cures' (a pill that eliminates autism completely) and 'treatments' (a pill that ameliorates severe difficulties only).[30] This is what Marta seems to allude to in Box 10 ('it's like managing it or getting rid of it, like, which, what does it do?', line 53), a point that was also raised in other FGDs:

[30] In the remainder of this chapter the term 'treatment' will be favoured over 'cure'. When the difference between the two meanings is relevant to the presentation of the results, it will be made clear in the writing.

> Mia: […] If they *[autistics]* had something that, like, could **lower their level of anxiety**, you know, and stop them from having sensory overload **rather than like eliminating their disorder entirely**… (FGD4)

Not only is the type of medication relevant to the way in which the targeted population and society may react to it; people's personal *experiences* of the condition also matter. In their final argument in favour of genomic research for the development of pharmacological treatments for autism participants asserted that, because autism takes different forms, acceptance and celebration of difference should not lead people to neglect the struggles that some autistics might encounter or ignore autistic people's desire to ameliorate their difficulties. In fact, some believed that the availability of treatments for ASD would likely be supported by autistic people:

> Rose: I think we should *[welcome genetic research to develop a pill for autism]* because **even if people, not everybody wants to take it, there will always be at least some people who do**, and it isn't really a matter of curing it or not, is kind of more an individual choice that you don't have to have autism, **you could be like 'ok, I don't want to take this drug', but equally you can.** (FGD2)

It is important to note that young people's endorsement of positive attitudes towards the development of ASD treatments was not grounded on the values underpinning the 'traditional' framework of biological citizenship, particularly citizens' responsibility for maximizing health by engaging in preventative or therapeutic interventions (Rose, 2007). As the emphasis on autonomy suggests, young people were in fact concerned that this form of responsibility could emerge from the availability of ASD treatments. Simultaneously, the value they placed on autonomy should not be interpreted as an indication that participants adopted a straightforward libertarian approach. On the contrary, their moral concerns show that in young people's accounts autonomy interests did not have an absolute value, as the next pages will demonstrate.

6.4.2. But there are some concerns

Participants' presumption in favour of the development of pharmacological treatments for autism was counterbalanced by several objections that can be clustered around the categories of 'coercion', 'identity', 'stigma' and 'diversity'.

Coercion

Young people were concerned that choices about curing autistic traits might be in part influenced by tacit forms of pressure. Because they valued the opportunity for self-determination that the availability of treatments would provide autistics, it is not surprising that coercive mechanisms that hinder such an opportunity were not welcomed. The analysis of this part of the FGDs identified two potential forms of tacit pressure, either from society or within one's family. For a society that sees autism as a problem, the availability of a cure could provide a 'quick fix' solution to address such problems rather than promoting a positive narrative around autism. In other words, as stakeholders have anticipated in the empirical studies reviewed in Chapter 3, treatments could become a 'convenient form of social control' (Russell et al., 2017, p. 5). The young people added that the development of pharmacological treatments could be used as a way to save public resources that would otherwise be spent on supporting autistic difficulties and this could lead to what the disability right literature calls the 'loss of support' argument[31] (Buchanan et al., 2000), i.e. a reduction of societal services and support for people with autism:

> Maureen: If sort of, like, what you were talking about earlier about **society kind of taking measures to accommodating people with autism, it may affect that** because if there is a pill that people can take then that could lead to people that might have taken precautions towards people with autism, they're just not doing it because they'd think '**well, they'll just take the pill and then it is not going to be a problem anymore because people can cure themselves if they need.** So it is not something that we need to think about because it's something that can be cured'. (FGD2)

[31] Please note, though, that in the disability literature loss of support seems to be understood as dependent on the reduction of the number of people with a certain disability due to the application of genomic technologies.

In most FGDs, participants assumed that it is autistic people who are confronted with the choice about ASD treatments, which may be because the thought experiment about the hypothetical pill featured two young people who discuss whether *they* would take the pill. Only a minority of groups (4 out of 12) reflected on whether their attitudes would change if the scenario was about parents making the decision on behalf of their autistic children. In these cases, participants discussed the impact that the availability of treatments may have on family relationships if autistic people experienced pressure from family members:

> Rose: I think maybe **family dynamics** *[could be affected by the availability of ASD treatments]* because for example, say a child didn't want to be cured of autism, you know, then they can grow up and **then now they are not comfortable with it. And, I don't know, their parent was quite keen on the idea.** And **this caused a rift,** it could actually be like more problematic than not having the drug in the first place. (FGD 2)

As Marta hints at when she mentions 'age of consent' (line 8), and as Adele expands in line 28 ('who are you doing it for?') in Box 10, participants added that family pressure would be particularly problematic if parents decided about ASD pharmacological treatments on behalf of their young children, who may lack decision-making capacities, and in doing so they were motivated by their own best interests, rather than their children's.

Differently from the argument about societal pressure, which has been applied to other psychiatric conditions (Kong and Singh, 2018), the concern about family pressure is more specific to autism, since ASD is often diagnosed in childhood. Childhood diagnosis may indeed raise 'questions about parental rights and duties which are often not mentioned in the philosophy of mental disorders' (Hens et al., 2019, p. 5), including questions about parental rights to cure their children's autism. However, this is not a new concern in the autism debate. Although neither cures nor pharmacological treatments for core autistic symptoms are currently available, self-advocates have been vocal about the harms caused by certain behavioural interventions their parents enrolled them in when they were young and unable to make their own choices. It could be objected that parents always make these decisions on behalf of their children, and that some in the neurodiversity

movement may be in favour of appropriate interventions. However, Chapter 3 has shown that what concerns critics about interventions such as ABA is that they aim at 'normalizing' autism, i.e. making autistics indistinguishable from their peers. As I will show below, similar arguments emerged from the group discussions in this study.

Identity

In Box 10 participants refer to the second concern that young people raised about treating autism, i.e. the impact that a pill might have on autistic people's identity, given that autism is such an integral part of it. A tension was highlighted between the positive outcome of ameliorating autistic people's difficulties and the risk of changing 'who they are'. In fact, in the quote below the same group comments that the former might be impossible without causing the latter, because it is difficult to separate autism from the person:

> Simon: **It's also, like, can you really separate autism from []**
>
> Marta: **The person.**
>
> Simon: **The person *[laughs]*, like?**
>
> Marta: What do they become if you take away the autism?
>
> Simon: Yeah, **what bits do you call autistic and what bits do you not.** And, like, because everyone has their weird, what's perceived as weird things that they do.
>
> Adele: Yeah.
>
> Simon: Like, are you going to take all of these away and are you just going to be – what, *[laughs]* **what's going to be left if** – like, how much is left? (FGD9)

Some autism scholars have similarly argued that autism cannot be considered as an 'appendage' (Sainsbury, 2009, quoted in Walsh, 2010, p. 522), but is instead 'constitutive of the person' or 'rather a condition than an effect of his or her conscious life' (Walsh, 2010, p. 522). This is why many people on the spectrum prefer the identity-first over the person-first language, which interestingly contrasts with recent efforts by health organizations to avoid disease-based, and potentially stigmatizing, labels in the context of somatic conditions (e.g. 'diabetics', 'asthmatics', etc.; see Ogden and Parkes, 2013).

As Walsh argued in her paper, so some participants in this study believed that the concern about identity is specific to discussions about autism. When asked to compare cures for ASD and for physical conditions, they indeed argued that what differentiates the two scenarios is that autism is 'really just their personality' (Sophie, FGD3). Although there is evidence that chronic conditions such as rheumatoid arthritis have a profound impact on people's identity and may cause a 'biographical disruption' (Bury, 1982), some sociologists have drawn a distinction between people who suffer from conditions that become problematic late in life, and those who have congenital conditions. While in most cases the former have experienced 'normal' or better functioning, and so 'there may be considerable interest, even a desperation, for cures', for the latter (e.g. those who identify with the Deaf culture, s2.2.2.2) 'impairment is their normality', and so they may not be enthusiastic about, or they may even oppose the development of cures (Shakespeare, 2014, p. 141).

Past bioethics work on psychiatric conditions, medications and ethics has focused on the impact that medications may have on the opportunity for individuals to develop their authentic self, which can be understood as 'self-discovery', i.e. listening to and being true to one's inner voice, or as 'self-creation', i.e. developing one's own life and creating oneself as one wishes to (Singh, 2012). In the present study, young people articulated a tension between 'coercion' and 'identity', which suggests that they were more concerned about authenticity as self-creation. As Ludovica argues below by comparing autism and ADHD, if cures that completely change 'who they are' are externally imposed on autistic people, this 'may not be what they want':

> Ludovica: I was going to say that if there ends up being a cure **it should always be an option, it should never be, like, 'oh you have autism, so you have to take this'**, cause I think with, like, some people with ADHD and stuff it's so often that they get given kind of just medications straight away **when actually quite a lot of the medication kind of completely changes who they are and that may not be what they want**, so I feel like always having the option of, if you want to, if it does end up being possible that there is a cure **it shouldn't be almost forced on people**. (FGD1)

This is consistent with findings presented in s5.6.1, whereby I argued that, perhaps due to their exposure to the recent public discourse around identity politics, young people valued self-

determination in the way in which autistics approach their own autism. Thus, although the identity argument was raised because autism has to do with people's personality, such argument does not seem to be about autistic identity *per se*, and so about the value of autism; rather it is about creating and choosing one's own self as one wishes to.

However, in a minority of cases participants seemed to be concerned about authenticity as 'self-discovery', when they observed that ASD cures may turn autistic people into 'what is perceived as a normal person' (Box 10, line 43), hence hindering their opportunity to be true to their inner self. This concern was brought to the publics' attention in 1993 when Jim Sinclair, founder of the self-advocate organization Autism Network International, gave the famous speech *Don't Mourn for Us* (Sinclair, 1993). He highlighted the tension between parents' wish that their children were not autistic and the awareness that their children would not exist without their autism – a tension often discussed in the broader literature about parenting a child with intellectual disabilities (Landsman, 2003). As seen above and in s3.4.2, this worry underpins the criticism of part of the autistic community against ABA and other interventions that, supposedly, aim at normalizing core ASD traits and are therefore grounded on a lack of recognition of the value of autism.

Notably, in contrast to what is reported by empirical studies with autistic people and other stakeholders (Reiff et al., 2015; Singh, 2016b), even when referring to autistic identity *per se*, the young people did not adopt genetic essentialist views, whereby autistics' genetic makeup is taken as the 'essence placeholder' of people on the spectrum (Medin and Ortony, 1989). Rather, identity was understood in terms of autistics' personality:

> Chloe: It *[an ASD cure]* just **takes people's personality away** from them […] It just, like, **if they did take the cure then it just takes people's personality away from them**. (FGD7)

In the quote blow, Cesar refers to autism as a 'lifestyle'. Although this might be because he is not a native speaker of English, his comment seems to confirm that the young people thought of autism more as linked to people's behaviour and conduct than to their biology:

> Cesar: I don't think it is necessary for a person to have a pill if he or she got diagnosed having autism in the age of, like, 30 or so, because they're already like getting used to their **lifestyles**. That person lived with it for 30 years without any severe problems so I don't think it's necessary for that kind of, yeah, to have the pill. (FGD10)

The lack of reference to genetic identity is in accordance with what was observed in Chapter 5, i.e. that genomic information had little relevance to the way in which young people from the general population thought of the nature of autism.

Only a few participants in two FGDs seemed to understand autistic identity as linked to people's biology, but through a neurobiological rather than genetic lens, hence using a language that reminds of the essentialist narrative of ASD developed by the neurodiversity movement. In FGD11, the group drew a distinction between autism, which they called 'mental disability', and other psychiatric conditions (e.g. depression), which they considered as 'mental illnesses', and they argued that, differently from the latter, autism should not and cannot be cured. This is because they saw autism as 'hardwired' (Ruby, FGD11) and so, as Scarlett stated, 'I don't think, from my perspective, that you would be able to actually rewire your brain or cure someone'. Leo added that 'short from going into someone's brain and rewiring their neurons', some traits such as ASD cannot be changed. In FGD7, two young people seemed to reduce autistics' personality to their brain:

> Noah: Yeah, it's quite dangerous meddling with such a large part of a **person's brain**. Because obviously autism affects just about everything they do and how they think, and **to eliminate that is, like, it's quite dangerous**.
>
> Participant: Yeah.
>
> Noah: Because it's, like, such a big part of how they think.
>
> Miriam: Yeah, like what Noah said, **taking that bit of the brain away from them doesn't make them who they are** and, as Noah said, **it would take their personality away from them**. Because that's, like, the best thing about all of us.

Stigma

Issues about normalizing autistic traits provide the opportunity to introduce the third category that emerged from the analysis of young people's discussion about treatments, i.e. 'stigma'. Either

spontaneously or when prompted, most participants argued that the mere availability of a cure for autism may increase ASD stigma, by implying that autism is 'something bad so it needs to be fixed' (Box 10, line 70). If society did not perceive autism as a disorder that should be cured, it would not make efforts to develop pharmacological treatments for it; in fact, autistics themselves would not want to change and so there would be no need for a cure:

> Cecilia: I think if you were more, like, accepted in society, if you had autism, **if people understood what it is more and, like, liked you for that, you wouldn't even want to change.** I think it's a fact that you just want to be like everyone else and fit in with everyone else. But **if people accepted you, you wouldn't mind having autism**. (FGD6)

Only one participant was sceptical about this argument. By making a comparison with people who have lost a limb but decide that they do not want a prosthetic leg, in the quote below Julia argues that the availability of a cure for autism may not have any effect on stigma attached to ASD; in fact, it may decrease it if autistic people showed that they did not want to be cured:

> Julia: I don't know if it's a different example, so if you lost a leg, for example, [unclear] there is prosthetic legs, that can be seen as a cure, but that cannot be said to be making it worse for people who don't choose to have a prosthetic leg because they just don't feel like they need one, so I think that's the same thing. **People don't think people who don't use the prosthetic leg are worse off than people**… do you get what I'm trying to say?
>
> Interviewer: You think that if you decide not to have a prosthetic leg you are stigmatized?
>
> Julia: No, no, **I'm saying you are *not* stigmatized**. So if you choose not to take the autism cure, I don't….
>
> Interviewer: You think there should be an option, but even if people decide not to take that option, that shouldn't be a reason to discriminate them.
>
> Julia: Yeah, and **I don't feel as if it will necessarily have that much of an impact on stigma; I think it can actually work in the opposite way.** If lots of people see that somebody said 'I don't want the cure', that shows that maybe it's not that bad.
>
> Interviewer: Mmm, I see.
>
> Julia: You know what I mean? With the leg thing, people think 'why would you not want a leg, how do you function?', then when they are, like, **'oh I'm fine, I can do everything I want to do', then that may change it around**. You know what I mean? People go to mountains and stuff who don't have legs, so it changes [unclear]. So I think it could happen the same.
>
> Ludovica: I think it is slightly harder in terms of the perceptions **when it's more to do with personalities rather than, like, a physical, with your leg is a physical injury and a physical disability**, it's a lot easier in that sense for people to kind of, for the stigma to be

> reduced and for people to have to, like, for people to understand it more, whereas I feel like with autism the understanding is a lot lower. (FGD1)

However, this argument encountered the resistance of participants in this and other groups, where I used it to challenge assumptions about stigma. In articulating their objection, young people recalled the distinction they had drawn between physical and psychiatric conditions, particularly how the latter impact on people's identity, or 'personality', as Ludovica phrases it above.

The argument about stigma shows that young people reflected on the background conditions that explain societal desire and efforts to develop pharmacological treatments for autism – and that they saw these emerging from stigmatizing assumptions about autism. These considerations do not feature in the views of parents interviewed about enrolling their children in ASD genomic research. While the opportunity for the development of better treatments is one of the main reasons why some parents would decide to enrol in research (see Johannessen et al., 2016), parents have not raised concerns about the negative judgements that their views may implicitly express about autism.

The category of stigma also emerged from the activity about priority-setting in autism research that I undertook with six out of 12 groups (s4.3.4.2). Participants' arguments suggest that most of them considered the reduction of stigma attached to ASD as one of the main goals of autism research; thus, this was one of the most important criteria for participants to decide how to prioritize the various research areas they were presented with. Similarly to the discussion about the neurodiversity argument, they thought stigma could be reduced by educating the publics about autistic traits and aetiology, which in turn could be achieved by investing in services that promote autistic people's inclusion in their communities, or involving society in discussions around sensitive issues related to ASD:

> Poppy: I think **'services'** should get at the top, like I mean, that's going to have such a big impact. Because there are going to be people who choose not to get the pill and they are still going to need help to get a job and stuff, and I think services also would interact with how… you know 'projects of community inclusion' *[Poppy refers to one example of services I gave to participants, s4.3.4.2]*, **that is going to help the society stigma**.

> Caroline: I was almost going to say services near… not near the bottom, but I would have valued the others more. **I'd say 'societal issues' near the top** because I think that's… I'd probably almost put that as number one because no matter… **even if you had these research into these different scientific areas, like there's still always going to be the stigma**, so I think that could be quite high. But I'd say services, yes, it is good to do that but if you had the other things first then you almost wouldn't need the…
>
> Alice: **Like if the stigma was gone**, then they might be able to…
>
> Poppy: I think… yeah, like I have the completely opposite, I think, yeah, 'projects on community inclusion', it's under services, and **that would just completely help eliminate the stigma** within, you know, that particular community, and 'employment', just getting autistic people employed that really helps their quality of life. And also together helps stigma because even if you eradicate the stigma, if they can't find a job, that's helping no one. (FGD 3)

The same group discussed whether research on autism genomics and autistic people's brains could be helpful in this sense, as it may reduce blame, in line with the slogan that 'mental illness is an illness like any others':

> Sophie: You don't need to see the structure of the brain to say 'oh this person has hypersensitivity towards…'
>
> Caroline: No, that's true, but if you could understand why that was happening and actually **prove solidly**, like, this is what is going on *[in autistics' brains]*, **then people would be more willing to be, like, 'oh it's not like just them being like a bit** *[weird]*'. (FGD3)

However, none of the groups ultimately thought that research on genomics or neuroscience should be prioritized. This was in part linked to concerns around the potential for this research area to lead to the elimination of autism:

> Mia: I think personally I'd just put them at the bottom because **I don't think you should try and eliminate autism completely**, I don't know, so… and I feel like that would lead to… **genetics, the whole purpose of that it's just like to eliminate autism** so… I think it is more important to just accept that there are autistic people and then just, you know, provide services and treatments for them. (FGD4)

Moreover, because investigations on autistic people's genetic makeup and brains are likely to produce benefits only in the long term, some young people argued that research should be devoted to improving services that can provide immediate support for autistic people:

> Joanne: I don't know, I just feel putting more time and money into the other areas *[rather than genetics]* will help. Well, **I think it's quicker to improve on those areas**. And I feel **the benefits are more direct**. (FGD8)
>
> Interviewer: Why everyone says that they want to put services at the top?
>
> Jasmine: Because **it's quite like an immediate demand**, like to help people.
>
> Mia: Yeah and I think kind of, like, just helps them integrate into society, like if it's employment opportunities and things like that, you know, because that would kind of have… yeah, you know, it just helps them I think to be normal.
>
> Evelyne: You can't just say, 'oh in 50 years we are going to have a cure, but hang on', *[giggling]* **because that's a bit late, isn't it**? (FGD 4)

These arguments resonate with the views of autistic people and other participants involved in past studies on research priority-setting (Frazier et al., 2018; Pellicano et al., 2014).

Societal diversity

Following considerations about identity and psychiatric stigma, the final category that emerged from data analysis concerned the implications that cures or treatments could have on 'societal diversity', which might be threatened by the development of a pill for autism. Across the FGDs, various reasons were proposed to support the value of a diverse and inclusive society, and these echoed young people's discussion about the neurodiversity argument. To give an example, 'curing' differences may lead to the elimination of the strengths associated with those differences and so the contribution that autistic people can make to society:

> Marta: And if you cured it then **you'd be getting rid of all of the, you know, the skills that they have, some of the [high abilities]** on the graph *[shown in the activity about neurodiversity]* and then **they wouldn't have the amazing ability in those specific areas, and they couldn't help society**. (FGD9)

Although this finding may once again reveal stereotypical assumptions about autistic special talents, it also supports views documented in studies with autistic people and their allies (Russell et al., 2017).

6.4.3. Summary

To recap, the analysis of the first activity revealed that young people acknowledged potentially positive implications of the availability of a hypothetical pill to cure or treat autism. These were phrased in terms of providing autistic people with the opportunity of making autonomous decisions about whether to be treated, as well as of enjoying the potential medical and psychological benefits that may be derived. The emphasis that participants placed on autonomy suggests that they did not consider autistics (or their parents) as having a responsibility, in the 'traditional' biological citizenship sense, for accessing pharmacological treatments to contribute to society's drive for health (Rose 2001).

However, this does not mean that they adopted a straightforward libertarian approach. On the contrary, autonomy interests were counterbalanced with a range of potential downsides of ASD treatments, which could impact either individuals or the broader society. Concerns revolved around the categories of 'coercion', 'identity', 'stigma' and 'diversity', and were informed by participants' dimensional understanding of autism as a heterogeneous condition characterized by traits lying on a spectrum, as well as their resistance against considering autism as necessarily a disorder. These considerations start to shake the portability of Rose and Novas's prescription to draw on biology to maximize health and minimize the risk of illness in the case of autism.

Similar concerns were raised in the discussions about prenatal genetic testing and genetic testing for early intervention purposes, although the next two sections will show that young people simultaneously developed new considerations.

6.5. Young people's attitudes towards preventing autism prenatally

The letter that I used to initiate the discussion about 'prenatal genetic testing' (i.e. PGD and PND, see Box 11) stimulated arguments in favour and against the adoption of testing for ASD. Data

analysis identified very similar arguments in discussions about PGD and PND; when differences relevant to the analysis emerged, they are highlighted below.

Box 11: Letter about prenatal genetic testing for autism (adapted from Moore, 2009)

When my eldest son, George, was born, I had no way of knowing that he had an invisible lifelong condition that, 19 years later, would keep him almost as dependent as a toddler. George is autistic. So is his brother Mark, born 22 months later. Knowledge of autism has galloped forward since their early days. Soon a prenatal test may be available, to indicate whether a foetus is likely to develop autism. I'm glad that test wasn't available to me.

My first pregnancy ended in a termination. A mid-term scan showed the baby had no limbs. Such a child, I thought, would have no option but to be a hero. I considered this an unfair burden and I ended his life, with great sorrow but without regret. It would seem logical, then, that I would have made the same decision about an autistic foetus. And yet there is no part of me that wishes away my sons' lives, or the life I have with them.

I have a third son, Jake, 10, who does not have autism. When I outlined the abortion debate, he was upset. 'George and Mark aren't sad about being autistic because they don't know they are,' he said. 'Anyway, what's wrong with being autistic?' Jake accepts the life he's always known. His brothers break his possessions, gobble up his chocolate, render the lavatory unfit for use, but he doesn't blame them. They're autistic; that's what they do.

Many articulate autistic people would argue that Jake's right; there's nothing intrinsically 'wrong' with being autistic, it's just that our non-autistic society does not meet their needs. Unlike my sons, who I believe have no perspective on their condition, the more able often suffer from awareness of their differences. I doubt that a prenatal test would be able to distinguish between severe autism and the 'high-functioning' kind, but in any case it's a mistake to think that life is easier for the more able. However, would able autists agree that their potential suffering should have been terminated before birth? I suspect not.

Most autists are physically healthy. A minority have epilepsy, and gut problems are quite common, but, for most, 'quality of life' depends on quality of care and understanding, appropriate education and a living environment that takes account of their sensory hypersensitivities. Such conditions are achievable – but at considerable cost to the carers, usually the parents. Not only is the caring hard work, but there's no cut-off point. My friends' children are away on gap years, starting university, finding partners. George and Mark are giant children and my responsibilities towards them will continue until I die.

A prenatal test without a termination would have prepared me a little for what lay ahead. But the thought that I might well have opted for termination makes me shudder. Abortion has always happened, and it always will. When the prenatal test is introduced, it will make no sense to decide that a mother can choose to terminate, say, a Down's baby, but is obliged to keep a potentially autistic one. It's inevitable that many will be terminated.

Autism often means sleeplessness, eating problems, self-harming, aggression, destructiveness, bizarre behaviour of all kinds. It can destroy marriages, and it certainly doesn't help your finances. I would never condemn a mother who decided that she couldn't cope with these possibilities. But autism also means unique perceptions, special talents, weird humour, a view of the world untainted by greed, envy, malice, vanity, ambition. A society that aims to remove all the variables that make human life so fascinatingly complex is not a society I want to live in.

Participants' negotiation of the different arguments is illustrated by the exchange in Box 12, where Matthew, Samuel, Ted, Luca, Gabriel and David from FGD12 discuss their attitudes towards preventing autism prenatally. In the present section, I will refer back to this passage to demonstrate that young people were in favour of the development of PGD and PND for autism based on considerations about autonomy and best interests, but they also raised many concerns that they articulated around the categories of 'coercion', 'stigma', 'diversity', 'limited clinical validity and utility' and 'being a good parent'. However, no concern was considered strong enough to argue that tests to prevent autism prenatally should not be made available.

Box 12: Young people weigh arguments in favour and against PND and PGD for autism

1M: What may be the arguments in favour or against Charlotte's *[the author of the letter]* view? And do you agree with or disagree with Charlotte's view and why?
3S: I mean, she says that every life is a good life and that you shouldn't abort an autistic person just because they have less of a good quality of life. But then, again, she did terminate her son that was born with no limbs, so it's contradicting itself.
6M: But I think it's quite fair where she says that she would never condemn a mother who decided she couldn't cope because I think if you don't think that you can do it, you don't think you can live with that or you don't think you can afford it, or you don't think []
9T: Give them a life they deserve.
10M: **Yes, you don't think you can give that person the life they deserve then I think you should be allowed to abort the child** because you won't be giving them quite as good a life as they deserve. **I don't think you should abort them just because you don't want an autistic child**. I think if you can cope then it's not right to abort them just because they're autistic.
14I: Why do you think it's not right?
15M: Because autistic people live… **most autistic people live fine in the world**.
16T: Yeah.
17M: And they just have maybe a slightly worse quality of life, but it's still a life. And I think you can't take that away.
19T: **Alex is always happy**.
20M: Yes, Alex is always happy and he's fine. He doesn't suffer because of it. So I think that's [unclear] that.
[…]
23L: The thing is this thing already happens. If there's some chance the baby will have some defect they might go abort it. It's just how it is.
25T: It depends on what []
26S: Yes.
27L: **I don't think autism should be treated differently, you know. It's just like any other trait or aspect of a person**. And if you're against it, basically it's like saying you're against abortion. Really, it's just that do you agree with the idea of abortion in general or not.
30I: So it doesn't matter what the condition is.
31L: I don't think it matters. **I just think it matters, again, what parents want, really**.
32I: So do you think that there should be an option anyway?

33L: **There should definitely be an option**.
34S: Yeah.
35M: If you can't support a baby, full stop, then you have it aborted. So if you can't support an autistic baby because you don't have the finances or whatever then you can have it aborted as well. I think it's just the same as that.
38L: Autism shouldn't be treated differently just because it's a [unclear].
39M: Yes. I think if we understood it better than we would, like, I think…
40T: **If they knew the severity before it was born, I think that'd be different**. If they said that your child is going to have very mild autism you're not really getting an abortion. But if they said it's going to be really severe, they're not going to be able to talk or hear or something then that's different, because with mild autism they can still…
44M: They can still live.
45T: I mean, like, Alex still does stuff really similar to what we do. Whereas when you can't speak and stuff that changes everything, it's a lot harder.
47I: What about Sam's case, like the profile that we read, this one?
48T: With Sam?
49M: I think if you knew they were going to have a life like that and they were going to be hitting themselves or hitting their parents, **I think you wouldn't want to live that kind of life yourself**. So I don't think… it would be like… so I think I'd probably get it aborted.
52T: That's hard because we've never been in that situation.
53G: **You don't know what level of autism they have. You just know that they're going to be autistic**.
55I: Yes, that's a good point.
56G: So if you get that test and you say 'oh, it's autistic' and then you'd be, like, 'oh, I'm going to terminate the baby', the baby can have very, very mild autism. It could be pretty much normal. []
59D: **And they wouldn't have 100% certainty. So you might be aborting a perfectly normal child**.
61S: The thing is, lots of babies have been aborted that would have grown up to have autism without the parents actually knowing. So there's other things they consider as well.
63I: And what about if it's not genetic testing on a foetus, but it's embryo selection? *[a brief explanation of this scenario follows]*
65T: **But then again, that's not going to be fool proof**.
66I: No, it wouldn't be. They would test them and see whether one has genes associated with autism, which means that the future child *might* have autism. But then you would select the embryo that doesn't carry the genes just to lower the risk.
69M: **I think that's different to the abortion**.
70I: Why is that?
71M: Because with abortion it's a definite []
72G: It's a definite action which you're taking.
73M: And you're killing it rather than... because when it's an embryo it's like it's alive but it's not like…
75I: Developed?
76M: Yes.
77D: **I think choosing an embryo is a lot less severe than choosing between actual born children or almost born**.
79S: Yes, because it hasn't been conceived yet. So it's a question of choosing whether to have it or not, and not whether to kill it or not, basically.
81L: **Planting a seed and then taking the seed back out again. That's not killing a tree**.
82S: **Or choosing whether to plant the seed**.

83D: It's like if you have a packet of seeds and you choose. Look, this one's the biggest or this one looks the greenest.
85S: Yes.
86L: **I don't think there's anything wrong with it**.
87D: Then when you put it in it doesn't guarantee it, but if you cut down a tree it's different.
88L: **It's a very personal decision but, again, the parents should *again* choose what they think is best**.
90G: Also, **if you're that scared of having an autistic child I don't think you're ready to be a parent, really**. **Being a parent is about going through it, taking what you get and just making the best of everything**. So if you're purposefully picking out a seed which isn't autistic, if you're going to that big of a measure **to have a perfect child, you don't deserve to really be a parent**.
95T: No one is ever ready to be a parent.
96G: Well, most people are and that's [unclear].
97T: No, it's not how it works.
98M: Because you can't ever be ready for that.
99T: Because you don't know what it's like to be a parent or to have a kid.
100M: And you don't know what it's like.
101G: What I mean are planned pregnancies, not accidental ones.
102T: It's the same thing.
103M: But surely this is proving that you are ready to be a parent because you're taking so much care with it that, like…
105I: But perhaps you're saying one of the virtues that parents should have is to accept whatever child you would have. Is that what you're saying?
107G: Yes, that's what I'm trying to say exactly. **Rather than 'picking up the best one'**. *[said in a funny tone]*

6.5.1. Welcoming the opportunity for reproductive freedom and best interests

In this activity most participants were in favour of PND (and eventually termination of pregnancy) and PGD for autism, and they made considerations about autonomy and best interests to support their attitudes. As participants repeatedly affirm in Box 12 ('I just think it matters, again, what parents want, really', line 31; 'There should definitely be an option', line 33), most young people across FGDs agreed that ultimately individuals or couples should have the opportunity to make their own reproductive choices:

> Interviewer: So when you say the choice is necessary, you mean it should be there?
>
> Joanne: Yes, **people should be able to**.
>
> Greta: **It should be there, yes**. What they do with that information is up to them, I guess, but **I definitely think that it should be an option**. (FGD8)

Thus, participants valued people's 'reproductive freedom' (Robertson, 2003), which in this case should be understood as people's right to decide whether or not to test their foetuses or embryos for genes associated with autism.

For some participants, respect for reproductive autonomy was particularly important in the context of autism due to the lack of adequate societal support for autistic people and their families. In the quote below, Maureen uses this observation to argue that testing and abortion may be more justifiable for ASD than for somatic diseases for which approved cures and supports are available:

> Maureen: I think it's different in the sense that maybe autism should be given an option *[PGD and PND for autism should be available]* but heart disease not, **because heart disease has a more kind of known cures**. So there is sort of… I think what she *[Charlotte]* says here is that there is no way out in that situation *[i.e. autism]* but I think if someone has **heart disease**... **So there are more cures** and that means that []
>
> Interviewer: Yeah, so that means that if there was more support, more societal support for autism, then maybe it would be…
>
> Maureen: **You wouldn't need an option**. (FGD2)

To a small minority of participants (two out of 56) who were explicitly against abortion and so did not support prenatal diagnosis for autism, others responded that, even if parents did not want to terminate a pregnancy, PND would promote their autonomy by providing them with the option to choose whether they want to prepare for their future child's risk of developing autism. The review of the autism literature (s3.5) has demonstrated that preparing for the future is one of the main reasons why parents value genomic information about their children's (susceptibility to) autism and, as will become clearer below, this argument often emerged from the discussion about EIs for autism.

Considerations about autonomy were intertwined with arguments about the future child's best interests. In line with the idea that there is a lack of societal support for autistic people and their families, and that children could be born with severe symptoms, participants argued that parents might decide that it would be best for the child not to be born, if they lacked financial and emotional

resources to offer them a good quality of life. In Box 12, the group phrases this in terms of not being able to give children the 'life they deserve' (line 9). Moreover, because parents generally want the best for their children, some couples might not want their children to suffer from severe autism, and so they might decide that the possibility of having a child with severe autism is a strong enough reason to terminate their foetus, as Julia seems to suggest in the quote below. Thus, the argument about best interests was grounded on considerations that any parent of a child with a health condition, or any parent in general, would make about their offspring's wellbeing:

> Julia: I think most **people want the best for their child**… um say if you have anything else, you want to make it better, **you want to make sure your child is happy**, and even though obviously people with autism are not, are quite different, **there's a possibility that they could be severely autistic** and very dependent, you know they can't do anything that everybody else can do; I don't know, I feel the same way. And if somebody would not want that for their child, they'd want to change it if they could… (FGD1)

Finally, when asked to compare PND and PGD for autism, only a small minority of participants argued that reproductive choices should not be about autism in the first place because there is nothing wrong with it, hence implying that there is no moral difference between PGD and PND in the context of autism:

> Poppy: I personally think that, like, that I would still find it *[PGD and embryo selection]* fundamentally decreasing the number of people with autism in the population because… and that in itself is kind of discriminating. **It'd just be better if, like, autism wasn't tested for in embryos**. (FGD3)
>
> Mary: I don't know how I feel about it *[PGD and embryo selection]* because I think in my mind is, like, **why would you reject something that could potentially become a life because of the perceived costs,** and I don't think that it's bad to decide that the costs will outweigh the benefits, but…
>
> Interviewer: So for you it doesn't make a difference if it's a foetus or an embryo… like the same, you would…
>
> Mary: No, I think it's… I feel like… **yeah, I feel like the argument lies within both**
>
> […]
>
> Mary: […] I think my argument is more like **embryo selection shouldn't be based around autism in the first place because I don't know if it should be regarded as a disability**. (FGD1)

The majority of groups considered PGD more morally acceptable than PND. This was not because of reasons related to autism *per se*, but because PGD happens at an earlier stage of life or, as many participants phrased it, embryos are 'just cells'. Similarly, in Box 12 (lines 81–87) participants use a botanic metaphor to draw a distinction between 'choosing to plant a seed' and 'killing a tree'. This is consistent with robust findings in the literature on stakeholders' attitudes towards prenatal tests for genetic conditions that, since early studies (Snowdon and Green, 1997), have demonstrated lay people's and professionals' preference for PGD over PND.

Based on these considerations, all groups argued that genetic tests to prevent autism prenatally should be made available, hence expressing more enthusiastic attitudes than the critical perspective held by the author of the letter (Box 11 above). However, their overall support did not prevent young people from raising a range of concerns which, as I will argue at the end of the next section, suggest that participants did not align with the risk-management model promoted by the 'traditional notion' of biological citizenship and genetic responsibility.

6.5.2. But there are some concerns

Young people's concerns about prenatal testing focused on 'coercion', 'stigma', 'diversity', 'limited clinical validity and utility' and 'being a good parent'.

Coercion

Similarly to their discussions about ASD treatments, participants worried that the mere availability of PND and PGD for autism could lead to 'societal pressure', or societal expectations that couples should prevent the birth of autistic children. These could manifest in tacit forms, such as negative judgements on those who decide to have an at-risk child despite a positive test result, which is at odds with the value that young people placed on autonomy in the context of reproductive choices:

> Poppy: Yeah, I worry that just society would just accept, not accept but, like, would **encourage people just to abort,** just… they won't be, like, people wouldn't even deeply think about it almost. Fair enough [unclear] 'no, I can't cope that baby is just not…' […] **people would be like 'why did you have him or her?'** (FGD3)

The tension between autonomous reproductive choices and tacit forms of (societal) coercion could arise as a result of the detection of any condition before birth; in Chapter 2 (s2.2.2.2) this observation allowed me to challenge Rose and Novas's assumption that biological citizens are always in a position to make free decisions about genomic information. However, according to the young people the case of autism is particularly problematic because autistics 'can live very happy and comfortable lives' (Rose, FGD2). In Box 12, participants give the example of their autistic friend, Alex, who 'is always happy and he's fine. He doesn't suffer because of it' (line 20). Seemingly, when asked to compare psychiatric and non-psychiatric conditions, participants in two FGDs differentiated between autism and life-threatening conditions. While testing to prevent the birth of a child with a life-threatening condition was less contested, the same did not apply to autism which 'is not like that, it's not a sentence', as Poppy from FGD3 put it.

Thus, it seems that at the core of the concern about tacit forms of pressure that could limit parents' opportunity to make their own decisions is young people's belief that autism should not be necessarily considered as a disorder or disability to be prevented. This introduces another set of concerns, which revolve around the category of stigma.

Stigma

The young people argued that as efforts to develop cures for autism could increase stigma attached to the condition, so the *mere availability* of PGD and PND might suggest that autism is such a problem that it should be prevented prenatally:

> Jasmine: I think **it makes it seem more as a disability** that people want to terminate their children because they might have it, it makes it, **it shows that society really does think of it as a disability and that something is really wrong with your child** because if you thought your child is going to be like normal and healthy then you would obviously, yeah. Do you know? (FGD4)

Along the same lines, the *actual implementation* of the interventions, i.e. people's decision to perform PND or PGD, might be grounded on stigmatizing assumptions about autism, particularly the idea that autism is a disorder rather than a difference:

> Ethan: In some cases I can understand it, like if the child was not going to have a good life, but if some parents were **to abort their child because of autism**, like, I personally wouldn't agree with that because **there's nothing wrong with them**, it's just they may be slightly worse at some things but slightly better at other things than other people. (FGD5)

Interestingly, in expressing their willingness to use genomic information for reproductive decisions, parents involved in past empirical studies have not always reflected on the potentially stigmatizing implications of their views. In one study about reproductive choices among mothers who carried the gene for Fragile X syndrome, participants acknowledged that their decision not to have more biological children might express negative judgements about their existing child with the condition (Raspberry and Skinner, 2011). In the only study on parents' attitudes towards prenatal tests for ASD, the issue of stigma was raised by a minority of participants, but only to argue that they would be willing to terminate an affected pregnancy precisely because of the stigmatizing views that their community holds towards autism (Chen et al., 2015; s3.5.1).

Societal diversity

Related to the category of stigma was young people's concern that the application of interventions meant to prevent autism prenatally would, as some participants put it, 'reduce neurodiversity'. This was considered problematic because of the value that young people attributed to a diverse society, as seen throughout this chapter. Not only did participants assert again the valuable contribution that autistic people make to society thanks to their special talents, they also commented on the scope that a diverse society has to encourage tolerance:

> Ludovica: I really like the last sentence *[in the letter]* about removing the unique perceptions and special talents and things **because I think the best functioning society is one that kind of has all the unique talents of everybody and celebrates them all and uses them all** to kind of… like, for the best. I think if there was the option of kind of, like,

> get rid of that and if we did get rid of what people see abnormal, I don't think it's very good for producing a high-functioning society. If that makes sense…
>
> Interviewer: So are you talking about like the importance of a diverse society?
>
> Ludovica: Yeah.
>
> Interviewer: And why is it important? Are you saying that because society altogether works better or because it is a better society?
>
> Ludovica: Yeah, **I think society works better** with... because if you just have one person's views and everybody has the same views then things won't get done because you need, like, to think outside the box **you need different perceptions and things to be able to kind of get the best outcomes of stuff**. And also I think as well as society working best, it creates the best society for, like, everybody around because **everybody is celebrated and respected**. (FGD1)

It was within this discussion that young people problematized the concept of normality. While in the first part of the FGDs they had affirmed that there is a continuum between autistics' and neurotypicals' behaviours (s5.5), in the second part the concept was challenged on a temporal dimension. One group argued that reducing neurodiversity is problematic because what is considered 'normal' nowadays (i.e. neurotypicals' traits) may not be in the future:

> Interviewer: And why do you think it would be problematic, let's say, if we reduced variety in society?
>
> Simon: I mean in general variety is just, it's nice to have this, like, just live in a world where there's loads of different people and things going on. **I don't think you should be able to say, 'OK, we're going to have a monoculture of, like, the perceived norm [of] people.'** I don't know.
>
> Marta: Yeah, the perceived, **what we perceive, yeah, changes over time**.
>
> Simon: Yeah.
>
> Marta: So what if we decide now that we want everybody to be, I don't know, Aryan and be really…
>
> Simon: Yeah, well, exactly.
>
> Marta: Or, like, skinny. Or else, like, **beauty standards, like, I don't know, 50 years ago it was a bit more all about curves and such like**. (FGD9)

Not only does this argument recall considerations about the contested validity of psychiatric diagnosis due to changes across context, time and culture (Rosenberg, 2006). That young people problematized the concept of normality confirms the suspicion, raised in Chapter 2, that it is not always possible to reach a consensus over the distinction between health and illness, perhaps

particularly in psychiatry. In turn, this challenges the obligation to maximize 'health' and minimize the risk of 'illness' with which the traditional notion of biological citizenship invests citizens.

Limited clinical validity and utility

A new argument was proposed about the 'limited clinical validity and utility' of tests (for a definition, see s2.2.2.2). Although participants did not explicitly use this terminology, they believed that reproductive choices about PND and PGD are more complicated in the context of autism due to its dimensional nature, which makes interpreting test results more difficult. As Marta says below, 'there's too many unknowns' in the case of autism. This is a consequence of the complexity of ASD aetiology, which implies that testing could generate a 'false positive'; and, as Adele argues, of the heterogeneity and spectrum nature of autistic traits, about which a prenatal test would provide little information in most cases. The group concludes that 'you cannot predict the future in these situations':

> Marta: Oh, OK. Um, I'm not sure. I, I think – I don't think, for autism, I don't think you should be allowed to abort based on a positive autism test result, because, for one, you don't know if that positive is – **it could be a false positive**. Um, **you don't know how, whether they're even going to get it, just because they have the gene**. You don't know how – like, **there's too many unknowns**. **It's not like that with Down's where you know for sure** and you're like, 'Oh no, I wouldn't be able to deal with that emotionally,' like, and I think that's fair enough, it's your decision. Um, but I don't think it's fair because, yeah, I don't know, **it's not certain enough**.
>
> Interviewer: Hmm, hmm.
>
> Adele: I mean she *[Charlotte]* even says, like, **it can be, like, I don't know, self-harming and eating problems, but then it could also be, like, talents, humour**.
>
> Marta: The good stuff, yeah.
>
> Adele: So it's like []…
>
> Marta: You're also getting rid of that.
>
> Adele: Again, what do you think it means? Because if they *[parents]* think obviously it's going to be like a negative type, then obviously they're going to result in probably, like, aborting because of the image that they have of the actual condition. But then, I mean obviously if you know enough about it, which no one yet does, then, like, that's fine.
>
> Marta: Yeah.
>
> Adele: But it's not – again, **it's like it's too umbrella-termed**. It's not – there's too many – there's not many specifics and there's not a checklist, like, you can't. So it's just – and I think also, **like you said earlier, you can be diagnosed with autism but then re-**

> **diagnosed with something else**, so therefore actually have **you aborted a child with autism with something completely different** which could have been easier to deal with or more complicated?
>
> Marta: Yeah, you don't – **you can't predict the future in these situations**. (FGD9)

This quote shows that young people reflected back on the subcategories of 'heterogeneity', 'spectrum' and 'multifactorial aetiology' discussed in the first part of the FGDs, and they reasoned about the impact that these have on the prevention of autism before birth. In this way, considerations about uncertainty emerged again in Part 2 of the FGDs, although not just in relation to young people's views of the nature of autism, but as a feature of the test results. Adele refers to the uncertainty around test results in relation to the contested diagnosis of autism: the acquisition of ASD genomic information before birth might lead parents to terminate a pregnancy, when in fact their child could be diagnosed with a different, perhaps more manageable, condition later in life.

Participants' arguments highlight the limited scope that couples have for applying risk-management strategies as the traditional framework of genetic responsibility would require good citizens to do. In particular, from their accounts it emerges that a circumscribed notion of genetic risk as necessarily providing actionable information does not fit the context of ASD genomics. This finding is in contrast with the study by Trottier et al. (2013), in which parents interviewed about their attitudes towards autism genomics research did not spontaneously reflect on the potential ambiguity of test results. When asked explicitly, they were optimistic that scientific progress will eventually allow the interpretation of currently uncertain information. The discrepancy might be because, differently from young people who have no special relation to autism, parents are likely to be personally and emotionally invested in the successful outcome of genomic research on autism, and could be vulnerable to scientists' optimistic discourse around the potential for autism genomics to produce useful information.

Of note, by putting young people's arguments under philosophical scrutiny it emerged that, like in Part 1 of the FGDs, so in these activities at times young people bracketed their dimensional

understanding, in order to propose more ‘comfortable’ dichotomous views (see Chapter 5). This happened in two ways. First, some young people seemed to overlook the complexity of ASD aetiology when they compared autism to Down syndrome (DS), to argue that the same arguments should apply to both in the context of reproductive choices, while in fact, as Marta alludes to in the quote above, risk calculations are more straightforward in the case of DS. This is because, although DS has a heterogeneous clinical presentation, it is caused by one extra chromosome 21, which has been detectable by karyotype techniques since the 1950s.

It is possible that some young people drew this comparison because in some groups I gave the example of DS to introduce the activity on prenatal tests and explain what these are used for. However, the analogy between the two conditions seemed to occur more consistently in FGDs with younger members, which is not surprising given that comments revealing deterministic understanding of inherited patterns in autism were more often expressed by younger participants (s5.4.2). For instance, in Box 12 Luca affirms that testing and abortion are already available for other conditions and because there is no difference between these and autism, prenatal testing should also be available for ASD (lines 27–31). Luca’s comment did not go uncontested, though; Ted reminded the group of the complexity of ASD by raising the concern that couples would not know about the severity of their future child’s autism when making reproductive choices (40–43), an argument that is then supported and expanded by other group members. Because in this FGD young people were all from the lower age range, this shows that not all younger participants expressed deterministic assumptions.

Second, some participants used their awareness of the small predictive value of PND and PGD to propose an argument that echoes the claim that ethicists call the ‘Great Beethoven Fallacy’ (Dawkins, 2008). As some had already anticipated in the discussion about the nature of autism (s5.6.2), PND and PGD could lead to preventing the birth of high-functioning autists, who could distinguish themselves in life for some great deeds. When doing so, young people seemed to

strategically emphasize one side of the ASD dimensionality (i.e. strengths) to support their attitudes against PGD and PND. However, as can be seen from the quote below, there were instances where participants remarked that they were aware that not all autistic people have special talents:

> Sophie: Also people who have made history are suspected of being autistic, **for example Turing, Da Vinci**, and they are also suspected of being autistic and quite severely so… to cope with it…
>
> Alice: They made such a big mark on the world.
>
> Sophie: And **I'm not saying that every child who is autistic is going to be that** but []
>
> Alice: It's a possibility. (FGD3)

Being a good parent

That young people did not share the idea that responsible couples have an obligation to 'live [their] life through acts of calculation and choice' (Rose, 2007, p. 147) became apparent in two FGDs. In these cases, participants explicitly commented on their conception of the good parent, who rather than aiming at the prevention of autism, should adhere to values such as loving and accepting diversity. One example is provided in Box 12 above. While commenting on embryo selection to prevent the birth of autistic children, Gabriel argues that being a parent means 'taking what you get and just making the best of everything' (lines 91–92), which resonates with the parental virtue that bioethicist Michael Sandel calls 'openness to the unbidden' (Sandel, 2007, p. 45). In Gabriel's account, if people are so worried about having an autistic child that they go through the trouble of embryo selection to have 'the perfect child', it means that they are not ready to be parents.

Another example is provided by the exchange below, where Scarlett affirms that most people would want to perform PND or PGD to have an 'easier' life, but she wonders whether the desire to prevent autism should be considered as 'normal' or 'weird'. Her view is shared by Ruby, who has a mentally disabled brother and during the FGD showed awareness of what it means to care for someone with severe difficulties and to see them struggling in life:

> Scarlett: […] But I think it's hard because it just depends on what you'd want. Part of me would feel cruel *[to do embryo selection]*. Why am I going to change it; if my child is autistic, then they are. **Why would I want to change that? Is that normal, is that not normal?** But if that's being offered to people then a lot of people are going to choose that option in life, aren't they? Because it probably would be easier. Living with a child with a disability is going to be harder. Everyone knows that […] **But it just, it seems weird at the same time**. I can't explain it. **Is that normal, doing that?** I know it's not but it's like genetically modifying the child, isn't it? Although it's your child, I don't know, **it is weird**, but is []
>
> Ruby: I get what you're saying. Personally, I'm just being honest, if I had to choose between having a disabled child or having a child with no disability, I would pick a child with no disability for the sake of the fact that I would want them to be happy. However, if I found out that I had a disabled child I would be fine. **I'm getting tested to see if I can have children now** and I'm not going to find out for a couple of years my results. **But even if I did find out I can have children and they would have disabilities, I would still have children** […] And I wouldn't care if they have autism because I think showing… I think nurturing a child can also change that. **When you breed good thoughts, good things happen. You want to still give that person a home, a loving family.** I would love to adopt a disabled child as well. **Just because someone was born differently I don't think that they should have a stigma attached to them** because they wouldn't want them. I personally think that if I had a disabled child then I have a disabled child. You can't change that, that's how it will be […] I would want them to have a life, have a chance to live their life. **And know what it's like to be in a world and be loved** and different stuff, no matter what disability they have. (FGD11)

Ruby has experiential knowledge of reproductive choices in the context of disability, because when her brother and his disabled girlfriend were expecting a child her family was confronted with the choice whether it would be best to terminate the pregnancy, which they eventually did not. In this quote, she affirms that she would access interventions to select an embryo with 'no disability'; however, if she could only have children who are born 'differently' she would want to have children anyway to provide them with 'a home, a loving family', and to show them 'what it's like to be in a world and be loved'.

Ruby's comment resonates with findings from studies on mothers' experiences of raising (mentally) disabled children. As Landsman has reported, although mothers would eliminate their children's difficulties if they could, so that their children would not struggle in life, their parenting experiences have taught them to appreciate forms of 'normality' that do not fit the expectations of the 'age of "perfect" babies' (Landsman, 1998, p. 92). Similarly, from the above quote it seems

that in young people's account good parents should be open to any child, 'no matter what disability they have', and love their children's difference. This finding aligns with the views of parents who understand the responsibility of good parents in the age of genetic technologies to be about accepting and honouring a child's specialness and difference (Raspberry and Skinner, 2011; Raz et al., 2019), rather than as an obligation to know about and act on their (future) children's genetic status. Thus, young people's argument adds evidence to the suspicion that the broad label of the 'biological' might not be applicable to every citizen, given that it seems to mask a wide range of (often opposing) experiences, values, interests, and ends.

A sub-finding within the category of 'being a good parent' emerged from those groups (six out of 12) that were asked to reflect on positive embryo selection, i.e. selection of embryos with higher chances of developing autism. Although they believed that parents should accept and appreciate any child, most young people opposed this option, primarily because they thought that a life with autism is often characterized by more hurdles. Thus, by selecting an embryo with higher chances of developing autism parents might act against the future child's best interests:

> Ludovica: **I feel that's more ethically wrong** in a way *[said in a very uncertain tone]*. Because **for people with autism there is more hurdles**, more struggles to overcome to live their life so you're kind of putting that onto your child. And I feel that, to me, that feels more morally wrong. But I don't know, it's hard.
>
> Interviewer: I saw you *[Carl]* were nodding.
>
> Carl: No, I just say that **autistic people do, kind of feel very isolated and… do face a lot of hurdles,** like she said, and challenges socially that they are not going to live a completely happy life…
>
> Interviewer: So you feel like they shouldn't be allowed to make this sort of…
>
> Carl: Not choosing them for the fact that they are autistic. (FGD1)

This suggests that in young people's accounts autonomy interests did not have an absolute value; rather, the future child's best interests seemed to trump considerations about parents' procreative liberty. In this sense, young people are not 'radical libertarians' but 'modern traditionalists', for

whom reproductive choices should be respected as long as they do not harm the future offspring (Robertson, 2003).

Although the scenario about positive embryo selection was meant to challenge the positive account of autism that young people had proposed in their discussions of prenatal testing for autism, this argument should not be seen as inconsistent with young people's earlier responses. It instead confirms that in most cases young people endorsed a dimensional way of thinking of autism, within which autistic strengths and the value of autistic lives can and should be highlighted without neglecting the difficulties that autistic people may experience.

6.5.3. Summary

The presentation of young people's attitudes towards prenatal testing for autism has shown that participants proposed two arguments in favour, which concern (future) autistic children's best interests and parents' reproductive autonomy. The appeal to best interests should not be interpreted as participants' alignment with the recommendation of the 'traditional framework' to optimize life outcomes. Young people used the principle of best interests within broader considerations around what a good parent would do (e.g. they would make sure to have enough financial and emotional resources to provide a good quality of life to their child); rather than because of a circumscribed preoccupation with managing a known genetic risk. Moreover, the emphasis that young people placed on reproductive autonomy suggests that participants did not believe that there is a moral obligation for parents to draw on genomic information to prevent autism prenatally.

This is confirmed by two observations. First, two groups were asked to comment on Savulescu's principle of procreative beneficence (2001), which affirms that reproducers should employ genetic technologies to select the child who is expected to have the 'best life' given the available information, including information about non-disease genes; if translated to the context of autism, this principle would support parents' moral responsibility not to have autistic children. The young

people disagreed with this option, and in the quote below Ludovica uses intensifiers to make her point stronger (‘definitely’, ‘completely’):

> Ludovica: **I definitely don’t think that there’s the argument that you have a moral responsibility to terminate your baby. I kind of completely disagree with that**… but then I guess it gets really difficult because you don’t want to force somebody into bringing in the child if they think they can’t support them and care for them and kind of look after them in the best way possible. (FGD1)

Second, young people raised many concerns about the use of prenatal tests for autism, which were informed by their dimensional account of ASD and uncertainty over its nature, particularly their resistance to the idea that autism should necessarily be considered as a disorder to be prevented. Participants were concerned that couple’s choices could be constrained by more or less explicit forms of societal pressure (‘coercion’), which might be grounded on and might promote negative assumptions about autism (‘stigma’), while simultaneously they valued diverse traits in society (‘societal diversity’). Moreover, as the category of ‘limited clinical utility and validity’ has shown, knowledge of the complexity of ASD aetiology made young people aware of the ‘many unknowns’ that characterize test results, which complicates risk calculations. Thus, some participants argued that ‘being a good parent’ means accepting and celebrating diversity.

As the discussion around pharmacological treatments indicated, young people’s arguments regarding prenatal testing question the assumption that a separation between health and illness can be agreed upon in autism genomics; and suggest that a linear link between genomic risk and action does not ring true in the case of autism. In other words, young people’s arguments did not seem to align with the recommendations of life optimization and risk management that characterize traditional biological citizenship projects.

6.6. Young people's attitudes towards genetic testing for early interventions

In the final activity young people reflected on whether genetic tests should be performed on very young children to assess their genetic susceptibility to autism for early intervention purposes. A minority of groups (three out of 12) were also asked about, or spontaneously discussed, the scenario of enrolling an asymptomatic at-risk child into EI programmes as a preventative measure. Because participants shifted from considerations about early testing to considerations about enrolling in EI programmes, and similar arguments applied to both scenarios, the categories that emerged from data analysis will be discussed together, unless otherwise specified.

As in the first two activities, no participant was strongly in favour of or against testing for EI purposes, but a variety of attitudes emerged. Young people perceived the knowledge of a child's genetic susceptibility to autism as empowering, as it may benefit the child's development and offer parents the opportunity to be prepared for what the future will hold. However, they also recognized the potential harms that may derive from such knowledge, both for children and their parents, and so discussed concerns that revolve around the categories of 'stigma', 'identity', 'medicalization of childhood', 'diversity', 'disrupting family dynamics' and 'limited clinical utility'. To illustrate these attitudes, throughout the next pages I will refer to the following passage from FGD1, in which Ludovica, Julia and Mary discuss genetic testing for early interventions for autism (Box 13).

Box 13: Young people weigh arguments in favour and against genetic testing for EIs for ASD

1I: […] So what do you think could be the benefits and harms *[of genetic testing early in life for EIs]* both medically speaking and psychologically speaking?
3L: I think **it can allow the parents to be more prepared** and to have, **to then provide more support**, because a lot of the times autism goes unnoticed and it's kind of, it's quite late the diagnosis, and so I guess **it may improve the quality of life of the child at an earlier age**, because they can get more support, their traits aren't just going to be dismissed and aren't just going to be ignored. It's more like **there is more support, there is more care for them**. So I guess that could be a benefit.
9I: Yeah and what could be instead the downside of that same argument, like the counterargument?
11J: **Constantly, like, judging them**, like you're just waiting for this and you're just… when are you going to be able to diagnose this person, it's going to happen. **You are going to have, to be thinking at all times, like, 'is this normal?' or 'is this just my child or is this the autistic traits?'** I think that's just… you're just, you know, **overanalysing your child** and then you end up with being even more um… he would, not over-diagnose autism… they would just think, **even though they are not what you would call autistics, they would end up treated as though they were autistic** and that could affect them later in life, because then **they feel that they have stigma attached to them**. You could have a lot more of that with the monitoring.
19L: **You could reduce their chances of them living more of a normal life**, in a way. Because you're kind of, like, constantly on their back.
21M: I guess at that point you're not treating them as a child or as a person**, you're treating them as someone who**, you know, if they haven't been diagnosed yet, **will eventually be diagnosed with autism**. And I feel like also the idea of monitoring, like your baby almost, I feel like **that takes away from the fact that autism is such an ambiguous thing so it's very individual**, so it wouldn't be fair to monitor certain behaviours because ultimately you won't be able to classify what could be an autistic trait and what's just normal behaviour for them.
27L: And if you'd be, like, labelling them too early on, because **once you've been labelled with it you're kind of stuck with it and you're stuck with, like, the negative connotations** that come with it, and I think if you kind of **allow a child to, like, grow at their own rate** and without kind of constantly… because I guess if you're monitoring them your kind of, like, maybe going to behavioural psychologist and stuff, **you are not allowing them live their life to the full.**
[…]
34I: Ok, so last question is, how would your response change if the child was enrolled in early interventions before showing any symptom of autism?
36M: **I disagree with it because you're not allowing them the opportunities to develop, say like, the more perceived beneficial aspects of being autistic**, if they are autistic.
38I: If they had already shown some symptoms would you be fine with enrolling them?
39M: If it was causing them distress and if it was, like, involving them being a harm, I would understand but if it's not, you know, proving to be a significant difficulty I wouldn't see the point in **putting them through something that makes it seem as it was an issue**.
42L: I think **it depends on the severity of their traits**, in terms of if they're causing themselves harm.

6.6.1. Intervening early could benefit children and promote parents' autonomy

When introduced to the scenario about genetic testing for early interventions, in most groups participants first thought of the positive implications that knowledge of a young child's genetic predisposition to ASD may offer (see Box 13), and they proposed arguments that often feature in the empirical literature investigating the reasons why parents value genomic information about their autistic children (s3.5). In particular, the young people seemed to view knowledge as empowering (see Ludovica's comments in lines 3–8 above). They associated testing with the opportunity for early diagnosis and support, which in turn could improve the child's quality of life; and with the option for parents to decide whether to know about their child's susceptibility, and therefore be prepared for what the future will hold. This argument had already been discussed in the activity about prenatal testing, and again young people did not present it as a moral obligation by which responsible parents should abide, but rather as a personal choice ('if you want to know'):

> Scarlett: I think **if you want to know** if your child is going to have a disability there's nothing wrong with that because **I think some people need to mentally prepare themselves** […] It is going to be a challenge but **if they can give themselves time to mentally prepare themselves**, prepare all their family, friends […] But it's not necessarily going to change everyone's views. It's just to prepare yourself. **That mental preparation of knowing what's going to come next**. (FGD11)

At first group members struggled to think of possible arguments against using the knowledge of a child's genetic predisposition to ASD for EI. This might be because they were not familiar with what EI programmes for autism entail. Therefore, as has been reported in other studies investigating children's attitudes towards gen-ethics issues (Bernhardt et al., 2003), regardless of their age some participants tended to focus more concretely on medical benefits and risks, and concluded that testing does not involve a 'dangerous' or 'harmful' procedure. As Samuel from group FGD12 put it: 'There's nothing wrong with that. That's just like checking if it's a boy or girl.' A similar point was raised about enrolling asymptomatic at-risk children in EI programmes:

Evelyne: Again, I don't know what would be in a programme, but **I can't imagine it would be harmful** to any child particularly, would it?

Interviewer: Harmful, you said?

Evelyne: No, no it wouldn't be, so if you did enrol your child in it with a 50% chance towards autism **it would probably be good for him anyway.**

Jasmine: mmm, yeah, like it's not going to have a negative effect, so **why would you not**?

Mia: Yeah and **it probably wouldn't even remember it**.

All laugh (FGD4)

Although direct comparative questions were not asked, the different reactions that young people initially had to the activity on pharmacological treatments and behavioural interventions for ASD may also be because of what bioethicists call 'bio-exceptionalism' (Horstkötter et al., 2014), from Green and Botkin's 'genetic exceptionalism' (2003). This is the tendency to focus on the potential implications of future biomedical measures while neglecting that the same concerns emerge from existing psychosocial measures (Manzini and Vears, 2018). However, when prompted to reflect on potential psychological and broader societal implications of testing for EI purposes, young people identified several concerns, which revolve around the categories of 'medicalization of childhood', 'diversity', 'stigma', 'identity', 'disrupting family dynamics' and 'limited clinical utility'.

6.6.2. But there are some concerns

Because in this discussion participants made similar observations to those raised in the activities about pharmacological treatments and prenatal genetic testing, in the present section I primarily focus on the new arguments or on those that were proposed with new nuances.

Stigma and identity

As Box 13 shows, participants formulated two concerns around 'stigma'. First, in line 41 Mary suggests that the option of testing and enrolling a child who may or may not have shown ASD symptoms in EIs could convey the idea that autism is 'an issue', and so increase societal stigma. Eloise from FGD10 had experiential knowledge of this kind of stigma-related issues. Because of

her family history of deafness, her parents had her tested for deafness as a child; when her deaf relatives found out, as she explained: 'I think they were quite offended. So I feel like you were saying, it might feel to some, like, it means that their life isn't equal to someone else's, I guess.'

Second, issues about stigma were intertwined with considerations about identity. As Ludovica affirms (Box 13, lines 27–29), labelling a child as autistic early in life may impact on their sense of self due to the negative connotations associated with autism. Other groups argued that labelling based on genetic susceptibility, which would occur before a behavioural diagnosis is possible, could become a self-fulfilling prophecy:

> Alice: Well, there's always the chances that knowing that they might have a risk **will make it, like, make the prophecy comes true**. You think they'll get it so you treat them in a certain way and they do get it. So you kind of push them unknowingly.
>
> […]
>
> Caroline: And then it's your point of earlier that it could cause them to start showing symptoms… not cause them to become autistic, but they might think 'oh…'
>
> Sophie: **They might have an identity crisis**.
>
> Alice: Yeah, they might think 'oh…'
>
> Interviewer: what do you mean?
>
> Alice: '…**I'm expected to…'**
>
> Sophie: Yeah.
>
> Interviewer: What do you mean by identity crisis?
>
> Sophie: **Well, if everyone is treating you a certain way maybe eventually you start** []…
>
> Alice: B**ecoming…**
>
> Sophie: **Becoming that…**
>
> Caroline: And it could really…
>
> Sophie: Like when everyone thinks you're stupid, you're going to think 'oh, maybe I am stupid, maybe, maybe I should just shut up and not…'
>
> Caroline: **It could really damage their self-esteem**. I am not sure how young they'd be when this would happen but if they start getting social skill classes they'd be like 'oh, I'm not…'
>
> Sophie: **If they are told that they're different they may not be able to grasp exactly how they are different**.
>
> Caroline: That will really damage…
>
> Sophie: **They'll feel separated from the rest of the children**. (FGD3)

This concern resonates with findings from the only survey that has investigated parents' attitudes towards testing to assess the risk for ASD in their younger undiagnosed children (Narcisa et al., 2013). While most parents expressed enthusiasm towards this option, a minority indicated that test results might attach a stigmatizing label to their children in their early years. Concerns around early labelling also emerged from young people's reflections over the risk of medicalizing childhood.

Medicalization of childhood and societal diversity

While young people believed that testing early in life could allow parents to be prepared for the future and benefit the child, their group exchanges revealed that they also worried about the potential for 'medicalizing childhood' (Conrad and Schneider, 1992), a concern that is often encountered in discussions about childhood psychiatric labels such as autism and ADHD (Singh and Wessely, 2015). As Ludovica remarks in the exchange above, 'you could reduce their chances of them *[children]* living more of a normal life' or 'you are not allowing them live their life to the full' (Box 13, line 19 and line 31). Julia adds that constantly monitoring the child might lead parents to 'over-analyse their child' (line 14), i.e. to interpret the child's behaviours through the lens of the ASD diagnosis. In other words, it seems that participants were concerned that genomic information might encourage genetic deterministic and essentialist tendencies in parents, which in turn could lead to the pathologization of certain behaviours potentially to the detriment of children's opportunity to enjoy their childhood and construct their sense of self as detached from their risk for autism.

These issues could apply to any behaviours considered 'abnormal' and have been flagged in critical accounts of psychiatry (Szasz, 1960), and more recently in ethical assessments of the use of genomic knowledge for EIs in mental health (Kong and Singh, 2018). However, participants also made an observation that is more specific to the case of ASD, when they argued that this form of monitoring could prevent children from developing the positive traits that are associated with autism:

> Noah: If they wanted to act upon it. Like, if they wanted to get them in, like, to try to stop it from developing, that may be, that may not be a good decision as it might, like, **the symptoms of the specific autistic individual might be positive**, and considering, um, **considering it can lead to great things, it may or may not be a good idea to cancel it out so early.** (FGD7)

Although Noah's reference to the 'great things' autism could lead to might once again reveal some stereotypical assumptions that grounded young people's views, this argument provides further evidence of the value that young people placed on diversity in light of their dimensional understanding of autistic traits and uncertainty over the nature of autism. This echoes the concern of those autistic adults who, because of their different conceptualization of autism compared with parents and autism practitioners, are uncomfortable with early autism research leading to earlier diagnosis or prevention of a diagnosis later in life, and instead favour research to better understand the unique development of people on the spectrum (Fletcher-Watson et al., 2017).

Disrupting family dynamics

A further issue that young people thought could potentially derive from testing for EI purposes is the 'disruption of family dynamics', which has already been reported in studies investigating the impact of newborn screening results for genetic disorders (Hewlett and Waisbren, 2006). Data analysis identified three ways in which this might happen. Knowing that a child has an increased genetic risk for autism might have a negative impact on parents, and consequently on the parent–child relationship. Parents' desire to prepare for the future should be weighed against the risk that awareness of their child's predisposition to autism may increase their anxiety. Anxiety is a concern that, as seen in s3.5, recurs in stakeholders' discussion about the implications of genomic information about autism in the empirical literature (see Johannessen et al., 2017; Tabor et al., 2011). However, in one group participants pointed out that knowledge early in a child's life could be preferential to finding out later on, when the child has developed autistic symptoms:

> Luca: **I doubt that it would be more serious than finding out that your child has autism in the first place, so I don't find anything wrong there again.**

> Gabriel: Yes, either you find out at this early stage and you start to try and help your child through it or you find out later on and it's a bit too late.
>
> Luca: **And then it's too late to help**. (FGD12)

Moreover, following the above concerns about stigma and medicalization of childhood, the young people argued that parents might start to treat their child as someone who will eventually develop autism rather than for who they are (see Mary's comment in Box 13, lines 21–23). This confirms young people's concern that parents could endorse genetic deterministic and essentialist tendencies, in line with the idea that biogenetic attributions trigger prognostic pessimism (s2.3.2). On this point, similarly to the activity on prenatal tests some comments suggested that young people endorsed a framework in which being a good parent means loving one's own children as they are, rather than being 'that kind of person' who only accepts the perfect child, as Gabriel argues below. In this sense, young people did not consider testing and enrolment in EIs as moral requirements, and they discussed alternative understandings of parental responsibility to those that the traditional framework of biological citizenship would propose in these contexts:

> Gabriel: I feel it's, is completely up to the mother what she decides to do because either way you're going to find out whether they're autistic or not. And **it doesn't affect them unless you're that kind of person**. **It's not going to affect how you love your child**, is it? **Unless you're that person who only loves the perfect child** *[said in a funny tone]*. And then again, you don't love it. It just depends really on the person. (FGD12)

Furthermore, a minority of groups (five out of 12) were introduced to the concept of 'baby sibs' studies (s3.3.1.2). Participants from these groups argued that these studies and the EIs linked to them could negatively impact the child's relationship with their older autistic siblings who, as a consequence of their parents' effort to prevent ASD in their younger brothers or sisters, might end up perceiving their condition as problematic. As Sally put it: 'The older sibling could take it as the parents don't want to have to deal with autism again […] And it could make the older sibling, um, feel angry.' (FGD7). This argument seems to confirm that the young people perceived the use of

ASD genomic information for EI purposes as potentially grounded on and as fostering stigmatizing attitudes.

Limited clinical utility

Finally, in some cases participants questioned the clinical utility of testing early in life. In this discussion, data analysis identified three main arguments that relate to the dimensional nature of autism. As Mary comments in Box 13, autism is 'such an ambiguous thing' and it is so 'individual' that knowing about a child's genetic susceptibility would provide little information about which behaviours should be seen as 'alarming' (lines 23–26). Additionally, because of the complexity of ASD genomics a positive test result would only provide a probability of the development of ASD, and so enrolling high-risk children in EIs would be 'a waste of time':

> Virginia: I think I would only actually enrol the child if they showed the signs of… the symptoms
>
> Rose: Yeah, me too.
>
> Virginia: If they didn't show any symptoms then, as you said, **if they don't actually have autism then it's just a waste of time**. (FGD2)

Jasmine from FGD4 was concerned that interventions based on a positive test result would be like 'stabbing in the dark' because in the absence of clear autistic behaviours it would be difficult to know what to intervene on:

> Jasmine: Yeah, like if they didn't have any symptoms or traits then you, **you're just, like, stabbing in the dark; they don't know what they are trying to intervene**, they are just kind of… and that might… I don't know if it could affect the child's personality or not but it potentially could and that might…

This is a concern that bioethicists have expressed in relation to very early and pre-emptive autism interventions (Singh, 2015).

6.6.3. Summary

While at first young people expressed positive attitudes towards testing for early interventions, because of the potential for promoting parental autonomy and children's beneficence, when prompted they described potential psychological harm to the individuals involved and broader societal implications. Thus, they referred to some concerns they had already expressed in the debates about ASD treatments and prenatal testing, which were grounded on their dimensional understanding of autism and uncertainty over its nature. These include the impact that the interventions could have on 'societal diversity', on 'stigma' associated with autism, and on the child's sense of self or 'identity'. Moreover, as they did for PGD and PND for autism, they discussed the 'limited clinical utility' of testing early in life, which is due to the heterogeneous traits of autism and its multifactorial aetiology.

Participants also raised two new arguments. First, monitoring children could lead to the pathologization of their behaviours, which limits children's opportunity for growing up as they wish to and for developing autistic strengths, and might in turn reduce societal diversity; thus, the concern about 'medicalization of childhood' seems to ultimately relate to the categories of identity and diversity that recurred in young people's discussions. Second, knowledge of a child's genetic susceptibility to autism could 'disrupt family relationships', particularly in the case of parents who aim to have a perfect child, or because older siblings with autism may perceive efforts to intervene early in autism risk as expressing negative judgements about them. Thus, this concern seems to ultimately relate to young people's reflections about being a good parent and their suspicion that the genetic-based interventions may be grounded on and may increase stigma attached to autism. In line with the first two activities, this section has demonstrated that, although arguing in favour of the availability of genetic tests early in life, young people did not consider parents as having a moral responsibility to draw on genomic information to intervene early on to mitigate autism risks.

6.7. Concluding remarks

This chapter has provided an account of young people's attitudes towards interventions that propose to use the knowledge of autism genomics to treat or prevent autism, or to intervene early in autism risk. Participants did not hold clear-cut positive or negative attitudes, but rather they weighed *prima facie* arguments in favour or against, which are summarized in Figure 12.

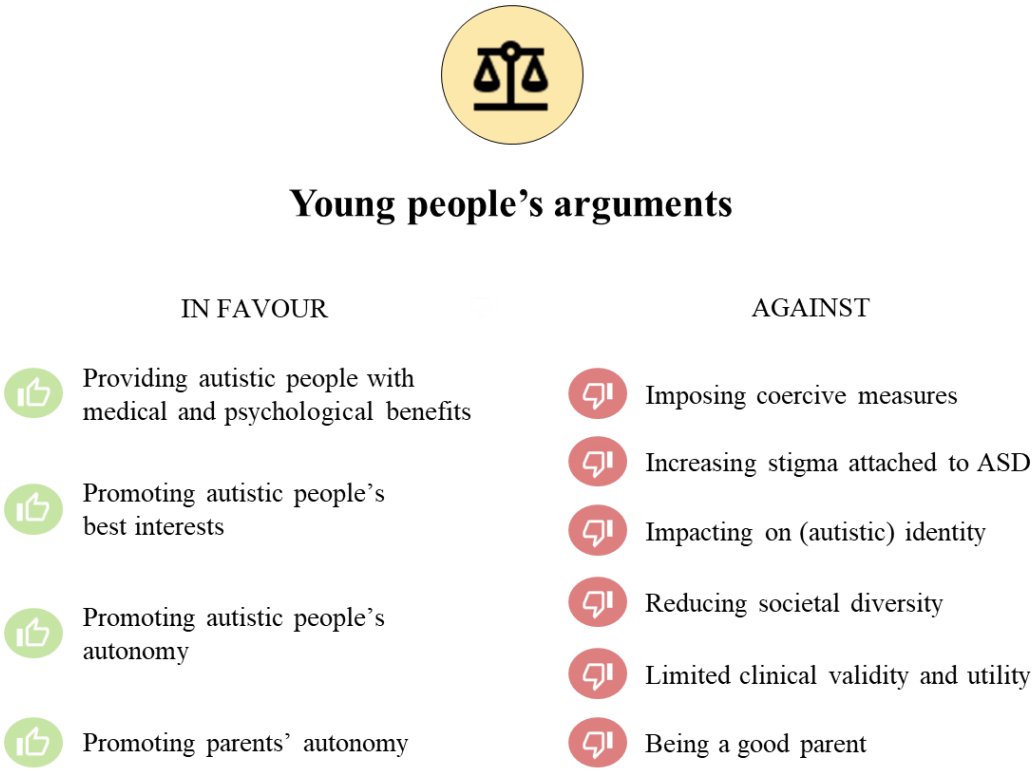


Figure 12: Young people's attitudes towards the interventions

Participants argued that the interventions for the prediction, prevention and treatment of ASD should be made available to allow autistics or their parents to make autonomous decisions on whether to access them. This is particularly important because pharmacological treatments and early behavioural interventions may offer medical and psychological benefits to people on the

spectrum. Further, some parents could decide that preventing autism prenatally may be best for the child, if they lacked the emotional and financial resources to raise a child who could have severe difficulties.

However, young people's endorsement of a dimensional understanding of autism and their uncertainty about its nature motivated a range of concerns towards using genomic information to prevent autism at different life stages. Given the value that young people placed on the opportunity for autistics and their parents to make their own decisions about the interventions, it is not surprising that participants worried about the risk that the availability of such interventions may lead to tacit forms of societal or family pressure to treat or prevent autism, or intervene early on it. Moreover, participants worried that ASD pharmacological and behavioural treatments could impact on people's identity in two ways. They could medicalize children's behaviours, and so prevent children from developing their sense of self as they wish to; in this sense, the concern about identity is a concern about self-creation, which reiterates the importance that young people attributed to self-determination. Second, they could impact *autistic* identity in particular, if medications were given to people who consider autism as an integral part of their identity, and early interventions hindered children's opportunity to develop positive traits associated with autism.

Thus, the implementation of the interventions could potentially reduce the number of autistic people, particularly if they aimed at eliminating autism, and so reduce societal (neuro)diversity and with it the contribution that autistic people provide to society. Participants argued that societal desire for the three interventions could be grounded on stigmatizing assumptions about autism, and that their mere availability could simultaneously increase stigma attached to ASD, by implying that autism is a 'bad' condition that should be prevented or eliminated.

In addition, because of the heterogeneity, spectrum nature and multifactorial aetiology of autism, the results of tests done prenatally or early in life would provide little information for people to make informed reproductive decisions or decisions about childcare. Finally, these concerns made

a minority of groups question whether parents should use genomic information about autism in the first place, as they argued that being a good parent means loving any child, regardless of their traits, and in fact appreciating their differences, rather than making efforts to draw on genomic knowledge so to have the 'perfect' child.

Two considerations are in order in the conclusion of this chapter. First, because participants proposed similar arguments across activities, they did not seem to see any strong normative difference between intervening at different life stages. Only in the discussions about PGD and PND did young people prefer the former because it intervenes at an earlier stage of development. However, their concerns (e.g. identity) acquired specific connotations when discussed in relation to different life stages, which indicates that participants attached slightly different normative nuances to the interventions by virtue of the timeline in which they are implemented. In this sense, the hypothesis I had developed when consulting the NeurOx YPAG, i.e. that the normatively distinctive feature of ethics discussions about autism genomics lies in interventions that are performed at different stages of life, was confirmed only to a certain extent.

Second, in line with the findings on participants' views of autism (Chapter 5), the discussions about the interventions confirmed that young people spontaneously adopted a dimensional way of thinking of autism, from which uncertainty about the nature of autism derives. This observation has allowed me to argue that participants' attitudes towards ASD genomics cast doubts on two assumptions underpinning Rose and Novas's biological citizenship, i.e. the 'alleged consensus over the distinction between health and illness' (assumption 1), and the 'linear relationship between genetic risk and action' (assumption 2). As the philosophical analysis has shown, only at times participants expressed dichotomous views. On these occasions, they supported deterministic views of ASD aetiology or stereotypical narratives about autism that allowed them to make more categorical and hence 'comfortable' arguments about the interventions. However, these did not take into account the complexity of the intersection between genomics and psychiatry.

In the next chapter, I will critically discuss young people's concerns to engage with the third assumption underpinning the traditional framework, i.e. the alleged conformity between societal obligations and biological citizens' choices and values. This will allow a broader reflection on what young people's attitudes reveal about the meaning that they attribute to the notion of biological citizenship in mental health, and why and how their perspective should be taken seriously.

Chapter 7. Discussion

7.1. An empirical account of ‘genomic citizenship in the making’

Having presented the theoretical framework of this thesis (Chapters 1 and 2), the case study and methodological design chosen for the empirical study undertaken within this research (Chapters 3 and 4) and the research findings (Chapter 5 and 6), the present chapter interprets and discusses these findings through the lens of the literatures reviewed in the background chapters. In this way, I will consider what young people’s arguments reveal about the meaning that they attribute to the notion of biological citizenship in mental health settings.

Building on Sabatello and Appelbaum’s writings (2016), I will call young people’s framework ‘genomic citizenship in the making’. However, I will move beyond Sabatello and Appelbaum’s approach, which seems to have interpreted the label ‘in the making’ in a developmental sense. The two scholars have focused on how to ‘raise’ young people so that they can eventually fulfil the role of the ‘traditional’ biological citizen, hence speculating on what children will *become* (s2.4.2). Because this study investigated children’s views and attitudes empirically, my focus is instead on children as genomic citizens in the *here and now*. I will argue that the empirical investigation showed that young people’s genomic citizenship in the making does not align with Rose and Novas’s framework, but is an alternative and equally valuable account. Thus, the ‘traditional notion’ is one of the possible understandings of biological citizenship, and should not be necessarily considered as the ultimate end goal of young people’s development as genomic citizens. This chapter concludes by reflecting on the theoretical and methodological limitations of the present study to discuss avenues for future research.

7.2. Summary of main findings

Before discussing what biological citizenship means to young people, in this section I summarize the main research findings. These include the results of the analysis of the qualitative data on young people's views of autism and their attitudes towards ASD genomic advances (s7.2.1) and the results of the philosophical analysis of young people's moral reasoning in psychiatric genomic debates (s7.2.2).

7.2.1. Young people's views of autism and attitudes towards the interventions

The analysis of young people's views of autism suggested that participants conceptualized ASD as a dimensional condition that is characterized by heterogeneous behaviours, a spectrum of severities and a multifactorial aetiology, although they attributed a higher degree of causation to genetics. Young people also expressed uncertainty over whether autism should be conceptualized as a disorder, disability, or difference for reasons grounded on their account of ASD as on a continuum of traits and severities. Interestingly, considerations about ASD genomics did not emerge from the discussions about the nature of autism; however, young people's multifactorial understanding of autism aetiology has likely contributed to create a complex picture of ASD that indirectly influenced participants' uncertainty.

When discussing the concept of neurodiversity young people agreed that society should do more to accommodate autistic traits, but they pointed out that some ASD traits are 'intrinsic' difficulties, which cannot be said to be caused by societal barriers; and that the needs of people with psychiatric conditions are likely to be more difficult to accommodate than those of people with non-psychiatric conditions. These considerations help to inform why participants expressed a presumption in favour of the interventions for the prediction, prevention and treatment of ASD based on the principles of autonomy, beneficence and best interests. Because some autistic difficulties and related social disadvantages cannot be alleviated by societal efforts, participants believed that the interventions

should be made available to allow autistic people or their parents' autonomous decision-making on whether to access them. Moreover, pharmacological treatments, and EIs following genetic testing, could provide medical and psychological benefits to people on the spectrum and parents could act in the best interests of their future child when opting for PGD or PND for autism.

However, young people argued that the availability of the interventions could hinder rather than promote autonomous decision-making, by imposing tacit forms of societal or family coercion to predict, prevent and treat autism; and that societal desire for the interventions could be grounded on and could foster stigmatizing assumptions about ASD. This was considered particularly problematic because, by virtue of their uncertainty about the nature of autism, participants did not consider ASD a 'bad' condition that should be necessarily predicted, prevented or treated. Moreover, ASD pharmacological and behavioural treatments could impact on autistic people's identity, and all three interventions could reduce societal (neuro)diversity, and hence the contribution that autistic people provide to society, thanks to the strengths that are part of the ASD spectrum. Finally, in light of their multifactorial understanding of ASD aetiology, young people remarked on the small predictive power of genomic information retrieved prenatally or early in life. Thus, a minority of participants questioned whether parents should use genomic information about autism in the first place, or whether they should instead love their children regardless of their condition, and in fact appreciate their differences.

This summary demonstrates that, because of their dimensional understanding of autism and uncertainty about its nature, participants did not hold clear-cut positive or negative attitudes towards the interventions, but proposed *prima facie* arguments in favour or against. However, the next section will show that there were occasions on which young people abandoned their dimensional account to adopt more comfortable dichotomous views. These allowed them to express more definitive attitudes, which were however grounded on stereotypical and deterministic assumptions about autism.

7.2.2. Young people's moral reasoning in psychiatric genomic debates

In this research, I put the empirical data under philosophical scrutiny as a methodological tool to distil the moral arguments underpinning young people's views and attitudes, by identifying where young people failed to meet the universal standards of reasoning in moral argumentation. As explained in s4.3.5, invalid arguments were discarded, because they made it difficult to understand what participants were trying to say; while valid but unsound arguments that neglected contingent features of the real world were interrogated to understand the reasoning underlying young people's comments. This was deemed essential to address potential scepticism about whether and why young people's insights should be taken seriously to drive deliberations in psychiatric genomic contexts. In this way, the philosophical analysis shed light on the way in which young people reason around ethical issues at the intersection between genomics and mental health (Figure 13).

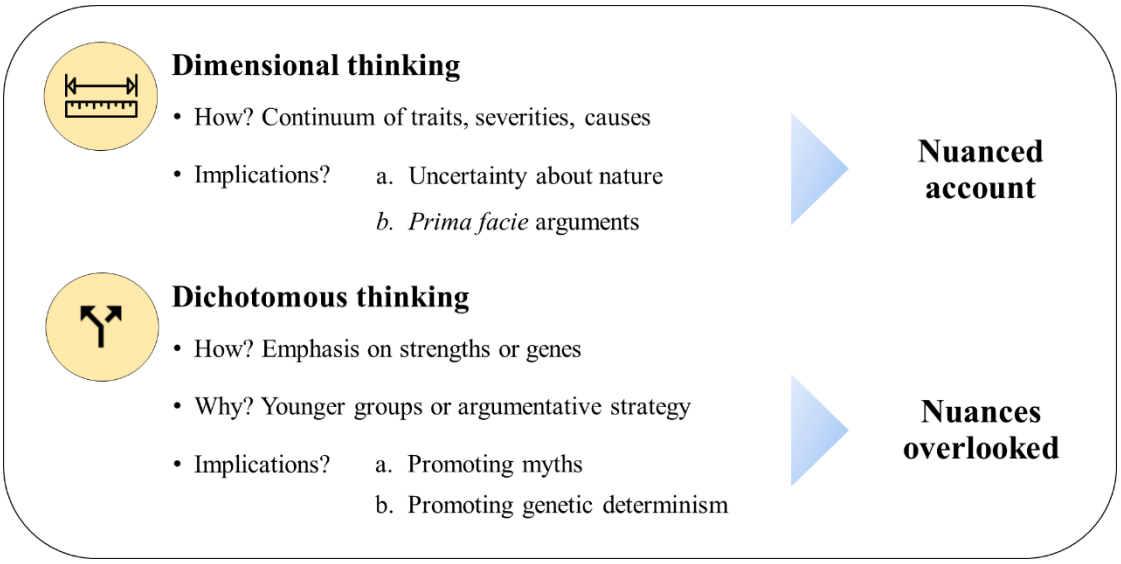


Figure 13: Young people's moral reasoning in psychiatric genomic debates

By placing young people's arguments under philosophical scrutiny it emerged that participants tended to spontaneously adopt a dimensional way of thinking of autism on a continuum of traits, severities and causes. Consequently, they expressed uncertainty about the nature of autism, which in turn led them to propose *prima facie* arguments in favour or against the interventions, rather than clear-cut positive or negative attitudes.

In the result chapters, I remarked on the importance of children's endorsement of dimensionality and uncertainty in this first investigation of the ethical issues that genomic citizens in the making identify in psychiatric genomic contexts. Indeed, this allowed participants to develop a nuanced account of ASD and of the implications of advances in ASD genomics, which contrasts with the scepticism about young people's capacity to contribute to discussions about issues of societal importance (Singh, 2016a; Singh and Keenan, 2010). By nuanced account, I mean that young people thought of ASD and the ethics of ASD genomics in a way that is consistent with the various layers of complexity that become apparent when genomics is applied to psychiatry (s2.3). These are the dimensionality of traits and severities of psychiatric conditions, which causes uncertainty around the value judgements that should be attached to them, and their complex aetiology, which makes risk calculations difficult.

However, at times young people adopted a more 'comfortable' dichotomous way of thinking of autism by placing undue emphasis on ASD strengths or on the causal role of genes in ASD development. Abandonment of dimensional reasoning in favour of dichotomies happened in two main circumstances. Groups with younger participants more consistently made comments that revealed they understood ASD patterns of inheritance in a deterministic way, for instance when they compared autism and Down syndrome in their discussion of prenatal tests (s6.5.2). Because, when challenged, even the younger participants showed they could reason around the complexity of autism genomics, this should not be interpreted as an age-related cognitive difficulty, but is more likely due to the fact that the biology curriculum in England does not cover complicated concepts

such as gene–environment interaction until Key Stage 4 (Year 11, Great Britain. Department for Education, 2013).

Regardless of age, dichotomous reasoning also seemed to be used as an argumentative strategy to 'serve context-dependent goals', as Condit would put it (2019, p. S27), particularly when participants wanted to promote a positive narrative about autism, back up their intuitions about the way in which autism develops, or support certain choices that intuitively they thought people should make about the interventions. To give an example, those young people who wanted to distance themselves from stigmatizing attitudes emphasized autistic special talents and argued that autism should not be prevented prenatally because autistics are capable of great deeds.

Various factors seem to have influenced these dichotomous beliefs and attitudes. Young people relied on their pre-existing knowledge, which is durable and unlikely to be easily replaced by new information provided within a research encounter (Weiner et al., 2017). This includes knowledge derived from ASD patterns of inheritance that they had seen in their own or their friends' families, which had inspired strong views on either genetics or environmental factors. Methodological issues might also have played a role. Participants may have responded to cues in my questions and explanations, which might have primed clear-cut responses, e.g. when I presented them with binary options (strengths vs difficulties, mild vs severe, genetics vs environment, disorder vs disability vs difference). Finally, the group setting might have elicited certain perspectives. As social psychologists have explained (Heine et al., 2019), impression-management motives that people consciously or subconsciously use to project a positive impression of themselves may shape the ideas that research participants endorse. Young people who wanted to present themselves as respectful of autistic individuals might have thought that accentuating ASD strengths was the socially desirable response to give in a group discussion with their peers. Nonetheless, this shows that they regarded respect for autistic people as an important value to discuss in debates about ASD.

Regardless of their specific context-dependent goals, in articulating dichotomous views young people ended up either promoting stereotypical views about autism, particularly myths about autistic special talents, which trivialize the difficulties that some autistics experience and are perceived as stigmatizing by parents and autistic people; or genetic deterministic views, which do not fit the current understanding of autism genomics and may lead to uninformed choices about the interventions. In both cases, they made arguments that neglect the complexities that emerge from the application of genomics to mental health.

Thus, the philosophical analysis of the empirical data highlighted that, while dimensional thinking led young people to co-construct nuanced accounts of ASD, when they adopted dichotomous thinking, participants proposed insights that did not take into account the nuances and peculiarities of the practical ethical context at stake. The implications of this way of reasoning about the ethics of psychiatric genomics on the conclusions of this thesis will be discussed below.

7.3. Interpretation and contextualization of the results

This section draws on the theoretical framework of this thesis to interpret the main findings. I will use young people's insights to make a threefold argument. First, the categories of dimensionality and uncertainty that emerged from the discussion of young people's views of ASD suggest that in most cases young people did not express genetic deterministic or essentialist views, which corroborates criticisms, including that of Rose and Novas, towards Lippman's 'geneticization' argument (1991). However, the ways in which young people tended to overlook considerations about ASD aetiology when they discussed the nature of autism cast doubts on Rose and Novas's argument that 'the biological' has become the driver for citizens' understanding of themselves and others. Thus, I will argue that Rose and Novas's traditional notion of biological citizenship is not a good *description* of the way young people as genomic citizens in the making think of health and illness in psychiatry.

Second, I will review the responsibilities that emerge from what, building on Sabatello and Appelbaum's writings (2016), I call young people's 'genomic citizenship in the making' to show that, on a *normative* level, children do not endorse the moral framework underpinning traditional biological citizenship projects. Their attitudes towards genomic-based interventions suggest that in discussions about the use of genomic information in psychiatric settings they prioritize a form of citizenship that does not align with Rose and Novas's ethics of active citizenship.

Third, I will argue that the responsibilities underpinning young people's 'genomic citizenship in the making' should be taken seriously because they derive from a way of thinking about autism that, as the philosophical analysis of the data has shown, takes into account the complexities that arise in the context of psychiatric genomics. In particular, because young people's genomic citizenship in the making derives from their understanding of autism as a dimensional condition, and their uncertainty about the value judgements that should be attached to it, I suggest that children's accounts could be applied beyond mental health to guide action in the use of genomic information about other conditions characterized by dimensionality and uncertainty. This section concludes by making the case for an expanded understanding of biological citizenship to include the ethical framework underpinning the notion of genomic citizenship in the making.

7.3.1. Beyond genomic '-isms', 'geneticization' and 'somatic individuality'

Data analysis indicated that participants used a neuro-genetic language to refer to ASD, and thought of some environmental factors as impacting the way in which autistics' brains develop in the early stages of life. However, this does not mean that they espoused genetic deterministic views. On the contrary, in s7.2.2 I explained that, with the exception of some younger participants, most co-constructed a dimensional and multifactorial account of ASD aetiology, and only strategically became determinists to serve context-dependent goals. This seems to confirm that genomic citizens in the making oscillate between genomic literacy and vulnerability to misleading information about

the power of genes (s2.4.2), and suggests that age plays a role in such vulnerability due to the way biology curricula are structured.

On the one hand, young people's limited endorsement of deterministic views of autism aetiology may reflect a broader shift in the publics' (perhaps specifically the younger generation's) understanding of health conditions from a genetic to a genomic lens. On the other hand, young people's insights are not necessarily an indication of genomic literacy, i.e. they do not demonstrate that participants had a scientifically accurate understanding of the complexity of ASD genomics. In this study, some participants did explicitly refer to the probabilistic nature of genomic information about autism, or to epigenetic processes, and hence they questioned the clinical validity and utility of genetic tests for autism. However, others made arguments that showed that they were unaware of genes' variable expressivity, incomplete penetrance, and dominant and recessive modes of inheritance, and this was particularly the case in the younger groups.

Thus, an alternative explanation for young people's limited endorsement of deterministic views may reside in the way lay people make sense of and navigate the discussion of complex phenomena. As Condit's work has demonstrated, in contrast to so-called 'experts', lay people are not trained to think of and interpret phenomena (e.g. autism) through the lenses of their specific discipline (e.g. genetics), but they store different causal features of human characteristics, to which they resort strategically (Condit, 2019). Indeed, evidence has shown that lay people tend to understand genes as probabilistic and amendable to manipulation via behavioural changes or technologies (Condit, 1999; Condit et al., 2001).

Notwithstanding the reasons for young people's non-deterministic account of ASD aetiology, it should be noted that such an understanding, coupled with their awareness of ASD multiple traits and severities, informed their complex account of autism; in turn, this explains their uncertainty in discussing the nature of autism. In this discussion, explicit considerations about ASD aetiology and groups' preference for a biological understanding of ASD did not feature (s5.6.2). This suggests

that participants did not ground their reflections of how autism and autistic people should be valued on their understanding of ASD aetiology.

Seemingly, young people thought of autism as fundamentally linked to people's identity; indeed, they questioned whether it is possible to treat ASD without changing the person (s6.4.2). In this sense, it could be argued that they held essentialist understandings, i.e. they thought of autism as defining the 'essence' of autistic people. However, only very few participants grounded this essentialist understanding on the idea that autism is biologically hardwired. Most young people did not discuss identity in terms of autistics' different genetic or biological makeup, but as their 'personality' (or 'lifestyle', s6.4.2). This, I argue, indicates that young people did not hold *genetic* essentialist attitudes which, according to the literature on biogenetic attributions and psychiatric stigma (s2.3.2), might have increased their stigmatizing assumptions about autistic people. On the contrary, young people made efforts to distance themselves from such assumptions.

If the above interpretations are correct, three arguments follow. First, this study seems to defuse bioethicists' and social scientists' concern that exposure to genetic words, concepts and explanations exacerbates socially harmful attitudes by making people genetic determinists and essentialists (Sabatello and Juengst, 2019); or at least this argument does not seem to apply to young people. It could be hypothesized that this is because participants in this study were not recruited due to their personal or family history of a genetic condition; thus, their encounter with genomic discourses that could have encouraged them to understand health conditions and identities, particularly autism and autistic identity, through a deterministic and essentialist lens was limited to their involvement in this research. Moreover, the young people were asked to reflect on others' identity, rather than their own. In contrast to the results of the present study, in Jennifer Singh's research (2016) there was evidence that the dominant geneticization of autism had been integrated in autistic adults' understanding of ASD and of their own subjectivities (s3.5.1).

However, bioethicists' and social scientists' concern is not meant to be confined to people's understanding of themselves, but to extend to their understanding of others. Moreover, it cannot be excluded that participants had been exposed to broader geneticizing discourses while searching for health-related information on the internet, as the account of genomic citizens in the making would suggest (Sabatello and Appelbaum, 2016), especially because some participants were members of Young People's Advisory Groups linked to hospitals or medical research centres. Furthermore, the present research is in keeping with evidence from Ilina Singh's interviews with children diagnosed with and treated for ADHD. Singh (2013) found that, although their daily lives are embedded within neuroscience discourses and practices, the children in her study have not constructed their subjectivities in a neurological sense.

The last example is not meant to suggest that the determining factor for the limited endorsement of genetic determinism and essentialism in this study was participants' age. In fact, the present research corroborates broader evidence, including evidence collected with adult participants, that has led scholars such as Rose and Novas to criticize Lippman's 'geneticization argument' (1991), according to which in geneticized societies citizens reduce differences between people to their genes (s2.2.1). This is why I sympathize to some extent with the argument that it is in fact bioethicists' and social scientists' 'ethics hype' (Caulfield, 2016) around advances in genomics that may be contributing to co-constructing geneticization (Weiner et al., 2017).

Second, from this discussion an important consideration should be derived about the literature on biogenetic attributions, essentialism and psychiatric stigma. Studies have shown that while reducing blaming attitudes, the increase in public acceptance of biological correlates of mental illness triggers essentialist thinking, which reinforces perceived dangerousness and unpredictability, the desire for social distance and pessimism about recovery. In turn, this leads to increased stigmatizing attitudes towards the mentally ill (s2.3.2). However, I argue that these

accounts have so far lacked a systematic reflection of what is intended by the publics' increased endorsement of biological and genetic explanations in psychiatry.

Although such a systematic analysis is beyond the scope of the present research, here I want to remark that increased acceptance of biogenetic attributions as causes of psychiatric conditions does not equal enhanced mental health literacy; on the contrary, if combined with little genomic literacy and an overestimation of the power of genes (Sabatello and Juengst, 2019), it may cause profound misunderstandings. As Read et al. have proposed (2006), perhaps what reinforces the perception of people with mental illness as naturally and so categorically different does not lie in presenting the causes of mental illness as biogenetic *per se*, but in constructing a disease model in psychiatry that neglects the dimensionality of mental disorders and so omits the complexity of psychiatric genomics. Conversely, findings from the present study suggest that when such a complexity is encouraged rather than omitted, essentialist and potentially stigmatizing views struggle to emerge. This explanation would support early evidence on people's attitudes towards different explanations of violent behaviour, which has shown that 'when people learned of a more accurate account for genetic influences on behaviour, they showed less essentialist reactions' (Heine et al., 2019, S25).

These first two considerations are not meant to deny the importance of ethicists' and social scientists' continued work on the ethical implications of the various genomic '-isms'. I acknowledge that misunderstandings and oversimplifications about genomics, perhaps particularly in such a sensitive area as mental health, are difficult to eradicate because of scientists' rhetorical and promissory discourses, the limited availability of adequate educational resources and lay people's and experts' psychological and cognitive mechanisms (Sabatello and Juengst, 2019). However, this study has demonstrated that lay people are capable of constructing or grasping nuanced accounts of complex human traits. Promoting such complexity could be one way of reducing genetic essentialist and stigmatizing attitudes among the publics, who may in fact 'already be more sophisticated about genetics, if not neurobiology, than is commonly thought' (Appelbaum,

2017, p. 16). In s7.4.2 below, I will reflect on the conditions and methodological choices that made this possible in the present research.

Finally, that young people referred to autism by using a neuro-genetic vocabulary and linked the condition to genes and brain development does not necessarily mean that they aligned with the profile of Rose and Novas's 'traditional' biological citizen. Although Rose and Novas have distanced themselves from Lippman's theory (s2.2.2), findings from the present research counter their wider assumptions about the impact that genomic advances have had on creating new ideas of personhood and subjectivity, particularly what they call the 'birth of the somatic individual' (Novas and Rose, 2000). In quotes such as the one below, I contest the idea that from biological languages, descriptions and definitions, citizens of the biomedical age necessarily derive value judgements about identity, and normative conclusions about how people should conduct themselves:

> *Like earlier languages – that of intelligence, or that of 'hormones' – these genetic languages render visible to others and to oneself aspects of human individuality that go beyond 'experience', not only making sense of it in new ways, but actually reorganizing it in a new way and according to new values about who we are, what we must do, and what we can hope for.*
>
> (Novas and Rose, 2000, p. 488)

The above discussion about young people's limited endorsement of genetic determinism and essentialism has indeed shown that biological presuppositions did not determine young people's understanding and moral appraisals of ASD. For instance, despite their preference for a biological narrative of autism, participants did not straightforwardly adopt a medical model, which conceptualizes autism as consisting of a range of biological deficits that should be predicted, prevented or treated (s3.4.1); nor did they endorse a neurodiversity approach, according to which autism is the result of a different brain 'wiring' that should be accepted and celebrated (s3.4.2).

It could be argued that the discrepancy between the present research and Rose and Novas's account is because autism is a multifactorial condition; thus, constructing a link between genetic

underpinnings and autistic subjectivity may be less obvious compared to the Mendelian disorders (e.g. Huntington disease) on which Rose and Novas have focused to draw broader conclusions about life in the biomedical age. However, similar suspicions about the tenability of Rose and Novas's notion of somatic individuality have been raised in studies focusing on conditions (e.g. familial hypercholesterolemia) that have different characteristics, including patterns of inheritance and lay understandings of them, and that are associated with different diagnostic technologies, availability of preventative measures and types of medical discourses and practices (Weiner, 2011).

By showing that Rose and Novas's theory of the birth of somatic individuals does not accurately reflect how young people think of autistic personhood and subjectivities, my findings are in agreement with the literature that challenges Rose and Novas's reductionist assumption that in western democracies there exists a hegemonic understanding of subjectivity in biological terms, that citizens have come to adhere to such a narrative, and that from this 'biological truth' they derive moral appraisals of themselves and others (s2.2.1). It also corroborates existing criticisms that question whether the broad label of the 'biological' is an accurate description of all citizens of current western democracies.[32]

In the next section, I will further criticize Rose and Novas's account to show that, as details about the biological underpinnings of autism have had little relevance to the ways in which participants understood autistic identity and subjectivity, so their ideas of being a responsible citizen in psychiatric genomic settings do not seem to be dictated by biological presuppositions. This may explain why young people did not subscribe to the risk-management and life-optimization model of the traditional form of biological citizenship. The values that young people promoted were not made to derive from a somatic idea of personhood and a normative framework that prescribes

[32] The same could be said about the label of 'genomic' citizens, unless one applies it to refer to citizens making choices about the use of genomic information, as I will do below.

people to ‘live [their] life through acts of calculation and choice’ (Rose, 2007, p. 147); rather, they proposed an ethical framework that values inclusivity of differences.

7.3.2. Young people’s genomic citizenship in the making

In Chapter 2, I argued that this thesis does not attribute a developmental connotation to the label ‘in the making’, in a way that may suggest that young people are deficient in biological forms of citizenship compared to adults. On the contrary, the label is a reminder of the *different* role that young people fulfil in society compared to that of traditional biological citizens, and serves as an incentive to investigate what we can *learn* from listening to young people’s views and attitudes at the intersection between genomics and mental health.

Young people are indeed interested in matters of societal relevance, including those related to mental health; in fact, their recent activism in local and global campaigns has shown that they are leading advocates for change in mental health (Booysen, 2019). Thus, it is important to discuss the concerns that they raise and the values they prioritize in this context, and to take these seriously by approaching them ‘as a basis for actual citizenship’ rather than for future citizenship only (Jans, 2004, p. 41), i.e. as having the potential to make a contribution to the ethics debate about psychiatric genomics.

In the following pages, I will use their views and attitudes to inform an understanding of biological citizenship in psychiatric settings from the perspective of young people. I will argue that what we can learn from young people’s contributions is that in making choices about psychiatric genomic information citizens should take into account the dimensionality of traits, severity and causes of mental health conditions, as well as the uncertainty that surrounds their nature. I will conclude that young people’s account of genomic citizenship in the making makes the expansion of the notion of biological citizenship a priority.

7.3.2.1. Beyond the ethics of active citizenship

As argued in the conclusion of Chapter 6, young people's moral attitudes towards using genomic information to predict, prevent and treat autism cast doubts on two assumptions on which Rose and Novas's account of biological citizenship is grounded. Participants' considerations about the limited clinical validity and utility of prenatal genetic tests or tests for EIs indicate that they did not assume that genetic knowledge is necessarily actionable or action-inducing (assumption 2, s2.2.3). Because of the multifactorial aetiology of autism, the prescription to make choices based on risk calculation is difficult to apply to a context where interpreting genomic information is becoming increasingly complicated.

Moreover, given the disagreement about how to conceptualize autism, young people were concerned about diversity. They argued that rather than predicting, preventing and treating autism, parents and society at large should accept, love and celebrate the diversity that it brings to society. This is particularly because their essentialist (albeit not *genetic* essentialist) understanding of autism meant that young people questioned whether interventions that might have a profound impact on autistics' identity should be used. Further, they were of the opinion that efforts to predict, prevent and treat autism may be grounded on, and may in turn foster, negative judgements about ASD, whereas they did not think that autism should necessarily be understood as a disorder to be eliminated. Thus, the obligation that, under the traditional account of biological citizenship, would prescribe responsible citizens to draw on genomic information to maximize 'health' and minimize the chances of 'illness' (assumption 1, s2.2.3) does not seem to apply well to the field of mental health, which challenges a strict dichotomy of 'normality' and 'pathology'.

By pulling these two points together, it emerges that participants did not adopt an approach based on Rose and Novas's ethics of *active* citizenship, and thus on the prescriptions of risk management and life optimization. On the contrary, young people's arguments allow us to derive a different understanding of biological citizenship in the context of ASD genomics, which I call 'genomic

citizenship in the making', and from the values underpinning their arguments a range of responsibilities that fall into this framework can be inferred.

In particular, in this study young people seem to have operated within a normative framework that promotes citizens' autonomous decisions about whether genomic-based interventions meant to predict, prevent and treat ASD should be used, and that moves beyond genetic essentialism and determinism to acknowledge the limited clinical validity and utility of such interventions. This framework also values inclusivity by encouraging considerations about the broader societal implications of choices in autism genomics, particularly in terms of stigma, and by supporting acceptance of and love for autistics' identity, and the celebration of the contribution that autistics' different traits bring to society (Figure 14).

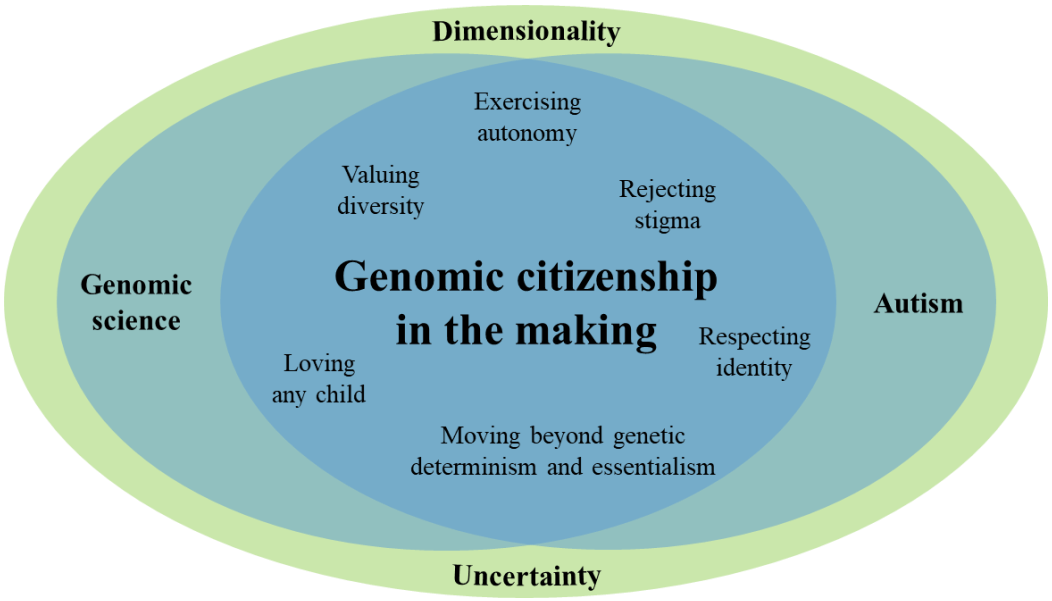


Figure 14: Young people's 'genomic citizenship in the making'

These considerations confirm the suspicion, raised in Chapter 2, that the traditional notion is too circumscribed not only to account for the various layers of complexity of the field of psychiatric genomics, i.e. the complex aetiology of mental conditions and the lack of agreement on whether they should be conceptualized as pathological, but also to accommodate young people's perspectives. In this way, the present study offers further evidence to challenge the third assumption underpinning Rose and Novas's account, namely that in the age of biomedicine citizens' choices and underpinning values always conform with societal obligations and expectations (s2.2.3). Indeed, young people add to those social groups who, by failing to match the profile of the neoliberal citizen-subject, promote alternative values to those of the normative framework underpinning biological citizenship projects.

In Chapter 2, I argued that these alternative normative frameworks make it evident that genetic responsibility is enacted within situationally specific values, interests and considerations, and that a notion of genetic responsibility limited to risk management and life optimization 'does not encapsulate the complexities that arise in practice' (Raspberry and Skinner, 2011, p. 427). In the next section, I will reflect on what is peculiar of young people's discussions about autism genomics, and thus what underpinning reasons have led young people to promote an alternative framework to Rose and Novas. In this way, I will explain why the values promoted in young people's account of biological citizenship should be taken seriously to guide action in psychiatric genomics and potentially beyond mental health.

7.3.2.2. A form of biological citizenship grounded on 'dimensionality' and 'uncertainty'

In the present research, young people were initially in favour of the interventions for the prediction, prevention and treatment of autism based on the principles of autonomy, beneficence and best interests. These principles are important for the analysis of the study results, as they show that young people did not propose clear-cut attitudes in favour or against the interventions, but *prima facie* arguments that need to be weighed against each other. However, these principles have little

to do with the practical ethical context at stake, i.e. they could have been applied to gen-ethics debates focused on any other condition.

It was when they started to focus on the complexities that arise from the application of genomic science to autism that young people proposed arguments revolving around the categories of identity, diversity, stigma, limited clinical validity and utility, and being a good parent. As the summary in s7.2.1 has explained, the values that participants defended emerged from the two underpinning categories of 'dimensionality' and 'uncertainty' that characterize the application of genomics to autism (Figure 14). In other words, young people's contributions suggest that in the field of autism genomics encapsulating the 'complexities that arise in practice', as Raspberry and Skinner (2011) would say, consists of endorsing dimensionality and uncertainty. This means that, when making decisions about the use of genomic information about autism, citizens should consider that risk calculations about autism may be more difficult than in the case of Mendelian disorders, because of the complexity of ASD aetiology, and that because of the dimensionality of ASD traits and severities, uncertainty surrounds the value judgements that should be attached to autism, and so disagreement exists on what constitutes 'health' (or 'normality') and 'illness' (or 'pathology') in the context of ASD.

Three considerations follow from this interpretation of the research findings. First, because any psychiatric condition is characterized by dimensionality and uncertainty (s2.3), I argue that the above considerations should be extended beyond autism genomics, to include psychiatric genomics more generally; autism is indeed a case study in this research. In other words, the account of genomic citizenship in the making illustrated in Figure 14 above is relevant to the broader field of mental health. Thus, young people's insights allow to derive an understanding of what biological citizenship means to young people in *psychiatric* (not just autism) genomic settings.

Second, I argue that young people's genomic citizenship in the making should guide citizens' choices about the use of genomic information in psychiatric contexts, i.e. it is a defensible

framework for psychiatric genomic settings, because the philosophical analysis of the data has shown that it derives from young people's nuanced way of thinking of autism that is mindful of the complexities arising at the intersection between genomics and mental health. In this sense, young people's framework is a 'better' fit for choices related to psychiatric genomic information compared to the traditional notion of biological citizenship, because it takes into account those nuances overlooked by Rose and Novas's more circumscribed account.

Third, although the categories of 'dimensionality' and 'uncertainty' emerged within a study focused on psychiatric (and autism) genomics, they are likely to characterize many other common complex conditions. Indeed, Chapter 2 has offered the example of non-psychiatric conditions (e.g. deafness and restricted growth) that present a range of symptoms, have a multifactorial aetiology and are on a continuum from normality to pathology, which causes disagreement on the value judgements that should be attached to them. Thus, although participants seemed to draw a strong distinction between psychiatric and somatic disorders (s6.3), the framework of 'genomic citizenship in the making' could apply beyond mental health settings to guide action in the use of genomic information about other conditions characterized by dimensionality and uncertainty.

In the next section, I will compare young people's genomic citizenship in the making with other autism stakeholders' perspectives, as documented in past empirical studies on advances in ASD genomics (s3.5), to further reflect on what we can learn from young peoples' contributions.

7.3.2.3. The case for an expanded notion of biological citizenship

Chapter 3 explained that the public debate around autism is still characterized by profound disagreements about how autism should be conceptualized, and whether and how genomic information should be used for the primary and secondary prevention of ASD. Although they expressed similar arguments in favour, the young people in this study seemed to focus more on the potentially negative implications of the interventions compared to parents in earlier research, some of whom enact a traditional form of biological citizenship by lobbying for ASD genomic research,

or seeking genetic testing for their children (see for instance Reiff et al., 2015; Singh, 2016b). As seen in s3.5, in some studies parents struggled to think of potential harms of, for instance, receiving individual genomics research results (Baret and Godard, 2011; Trottier et al., 2013); when they did discuss their concerns about ASD genomics, they tended to focus primarily on potential harms to their children and families, rather than considering broader societal implications (e.g. reducing societal diversity).

This suggests that the young people may be less enthusiastic about advances in autism genomics compared to parents of autistic people, which may be because the young people in this study have been raised in the 21st century, a time of intense activism by autistic adults and their allies, aimed at promoting a positive imaginary of ASD (Fitzgerald, 2012). Thus, this finding may be indicative of a generational shift in sensitivity, or positivity, towards autism.

However, in light of the evidence of persistent stigmatizing attitudes and bullying towards autistic children by their peers (see White et al., 2020), there might be another explanation for young people's emphasis on potentially negative implications of the use of ASD genomic information, which relates to children's 'special' role in society. Not only do children tend to have capacities (e.g. being more imaginative) that are qualitatively different from those of adults, as argued in s2.4.1; moral philosophers have also theorized that one of the 'goods' that makes childhood intrinsically valuable is that children have relatively fewer responsibilities and more free time compared to adults (Brennan, 2014; Gheaus, 2015). This could explain why young people's attitudes did not align with those of parents who, although not interested in ASD genomics *per se*, engage with genomic science more pragmatically, e.g. as a way to receive a free ASD diagnosis for their children or access (better) ASD services (see Singh, 2014; Tabor et al., 2011).

Because participants in this study were young and most did not have direct experience of autism, it is likely that they had fewer (or no) caring responsibilities towards autistic people compared to the populations involved in earlier empirical studies (i.e. parents, autism professionals and autistic

adults). Thus, because they were not involved in the micropolitics of making choices about ASD genomic information in the context of family relationships, they had the 'luxury' of reflecting on issues of broader societal implications, including identity, stigma and diversity, in the FGDs. In this way, young people's concerns diverged from the preoccupation with more pragmatic interests relevant to family experience that is distinctive of most socio-empirical studies on genetic responsibility, as identified in a recent systematic review (Leefmann et al., 2017).

However, young people did not completely reject the genomic-based interventions discussed in the FGDs. This means that they also disagreed with the most radical neurodiversity activists and autistic adults who oppose the use of genomic knowledge to prevent and cure autism, e.g. through early diagnosis, medications, or behavioural interventions to prevent a diagnosis later in life (Fletcher-Watson et al., 2017; Frazier et al., 2018; Pellicano et al., 2014; Russell et al., 2017). Because they believed that autistics' social disadvantage cannot be alleviated exclusively by societal efforts, young people expressed a presumption in favour of the interventions, based on the principles of beneficence, best interests and autonomy.

The above considerations suggest that, thanks to their position in society as younger members of the general public, young people co-constructed a complex account of autism that is inclusive of different options about the use of ASD genomic information. Except for those who had siblings on the spectrum, the study participants did not have a direct and personal involvement in the research topic, and so they might have not been interested in 'defending' strongly polarized views and attitudes. As argued in the results chapters, young people's openness to different options might have also been influenced by their exposure to recent discourses around identity politics in relation to people's gender, race and religion (see Bragg et al., 2018). Thus, participants promoted a normative framework that, although only proposing a range of responsibilities rather than prescribing a specific course of action (or perhaps precisely because of this), rests in between the polarized perspectives that still characterize the public debate about ASD. In this way, young

people's insights enrich the debate around advances in autism genomics by contributing to recent efforts to identify a common ground between different models of autism.

I do not intend to argue that young people's genomic citizenship in the making is the 'right' form of citizenship that should be exercised at the intersection between genomics and mental health, supporting the 'right' responsibilities that citizens should enact, and based on the 'right' way of conceptualizing autism and other psychiatric conditions. This would imply that parents' active citizenship and autistics' opposition to some applications of genomics science promote 'wrong' citizenship claims. As has been noted (Leefmann et al., 2017; Raspberry and Skinner, 2011), the notion of genetic responsibility and its underpinning values take (and rightly so) different meanings depending on the role that one fulfils in society, the characteristics of the disorder at stake, and moral agents' experience of such disorder, which give rise to different, yet legitimate, needs, priorities, and interests. Some bioethicists would argue that this is because moral reasoning inevitably has a subjective component deriving from the agent's distinctive standpoint towards the moral issues at stake (Ives and Dunn, 2010).

In this sense, the framework of genomic citizenship in the making does not have absolute moral value, i.e. the responsibilities that it promotes are not the only morally relevant aspects when it comes to choices about the use of genomic information in the context of autism, or any other condition characterized by dimensionality and uncertainty, as seen above. This further confirms that Rose and Novas's traditional notion of biological citizenship should not necessarily be considered as wrong, and so it should not be *substituted* by young people's account. Instead, like other scholarly work (see Raspberry and Skinner, 2011), this thesis makes the case for an *expanded* understanding of biological citizenship to include the ethical framework underpinning the notion of genomic citizenship in the making.

In the remainder of this chapter, I will reflect on the theoretical and methodological limitations of the present study to highlight opportunities for future research.

7.4. Limitations and directions for future research

7.4.1. Theoretical considerations

The theoretical limitations of this thesis are due to the interdisciplinary nature of the research, which required me to make a number of choices about the way the research topic was addressed. This is, indeed, the first study to investigate young people's moral attitudes towards psychiatric genomic advances, and to do so through the lens of the literature on biological citizenship. This means that the theoretical framework of this thesis is informed by various academic traditions and underpinning ethical issues, which prior to the present research had yet to be incorporated into a single empirical bioethics project. Thus, to accommodate the novelty of the interdisciplinary ground that this thesis covers, the different approaches that have been included in this research could not be reviewed exhaustively.

Moreover, as explained in section 4.2.3, this research was part of the *Citizens* project, which commits to enriching policy-making and practice at the intersection between psychiatry and innovation through youth engagement. When I started this research, I was convinced of the importance of creating the space for young people to express their moral concerns about psychiatric genomics, because today's children will be faced with ethical questions arising from the progress currently being made in this field. However, the novelty of the research topic meant that I was primarily interested in testing the traditional notion of biological citizenship in the context of mental health and in relation to young people. As such there was no space to investigate potential policy and practical implications of the research findings, on which this thesis therefore remains largely silent.

Despite the anticipatory, and to some extent speculative, nature of the research topic, which focused on interventions that are not yet standard applications of genomic science in ASD care, this study has allowed young people's sensitivity towards ethical issues in mental health to emerge. Thus,

future research should use the findings to develop, together with young people, an ethically robust agenda for psychiatric genomics research and its translational applications, so that these are mindful of young people's concerns and values. For instance, the responsibilities that fall into the account of genomic citizenship in the making could inform guidelines to facilitate communication between professionals and families in counselling sessions. Psychiatric genomic counselling is indeed becoming increasingly popular and already integrates information provision-based care, focused on explaining genetic risk for mental illness and opportunities for genetic testing, within psychotherapeutically oriented care that supports families to manage their expectations, feelings, hopes and values related to psychiatric genomic information (Hoang et al., 2018b; Inglis et al., 2015).

7.4.2. Methodological considerations

A number of potential biases introduced in the data due to the limitations of the study sample and methodological choices should also be thoroughly considered. First, in this study I recruited a relatively small number of participants via convenience sampling, which is a non-probability form of sampling that does not allow to generalize the results to a larger population. However, as explained in s4.3.2.2, this is an acceptable sampling strategy in exploratory studies that do not aim at generalization (Pearlman and Starks, 2010). Second, in this research two FGDs included two and three participants respectively (see s4.3.4.2). Because in this study interaction between group members played a particularly important role in allowing participants' arguments to emerge, one could wonder how this was achieved in such small groups. In addition to my active role as a facilitator, with which I aimed at ensuring the same kind of dynamic interaction that characterised the other FGDs, in these cases the tasks in which I involved participants proved to be particularly helpful, as they offered additional viewpoints with which the young people were encouraged to engage.

Third, the children who participated in this research might have not been representative of the average young person with no 'special' relation to autism. Some participants had close relatives on the spectrum, and so they might have been more knowledgeable about autism than others. In three groups (all or some) participants were members of YPAGs, who are used to being asked to express their opinions about health-related topics; and young people who participated in this study while already undertaking other activities within the NEUROSEC team were trained in discussing ethical concepts in psychiatric genomics. Moreover, because this study required a significant time commitment for a small incentive (s4.3.3), it is possible that only those who were already interested in autism volunteered. Therefore, the views and attitudes identified might be more sophisticated than if a 'purely' general population had been involved and they are not generalizable beyond this research, although the requirement of generalizability does not fall under the scope of qualitative research.

Moreover, participants were equally distributed between the two age ranges (i.e. 50% were 13–15 years old), but the majority were females (69.7%) and most young people were White British (60.7%, see s4.3.4.1). In addition, although participants recruited from schools, who account for the majority of young people in this study, attended a mix of state and private schools, I could not calculate participants' socioeconomic status (SES) due to an inability to obtain reliable data from young people, as others have noted (Svedberg et al., 2016). In the iterative process of data collection and analysis, I carefully reflected on the risk of introducing potential biases due to the strong preponderance of females in the focus groups, and I considered the option of purposively recruiting more males. However, data analysis identified differences in responses only due to age, while similar views and arguments were proposed by female and male participants. Although the research topic does not straightforwardly call attention to any other demographic feature that could play a role in children's views of ASD and attitudes towards the interventions, future research should aim at recruiting a more diverse sample and assess whether participants' values might change depending

on their demographic characteristics, including their SES as calculated by more child-friendly measures such as the Family Affluence Scale (Hartley et al., 2016).

Furthermore, this study did not seek to recruit participants on the autism spectrum, but no criterion was introduced to exclude autistic young people (s4.3.2.1). However, none of the children who eventually participated had received an ASD diagnosis. Thus, the present research, which has contributed to move the literature on biological citizenship forward by involving young people from the general population, lays the groundwork for future research to address the same topic from the perspective of young people on the spectrum, whose views have yet to be investigated. Such research should consider what this population's views and attitudes tell us about the meaning that they attribute to biological citizenship in autism genomic contexts. It would be interesting to assess whether and why their perspectives might differ from those of adult self-advocates, especially those with more radical views, whose opposition to genomic research might have been contextually dependent on a time, prior to their and their allies' intense activism, in which the dominant view of autism was that of parents in desperate search for an ASD cure.

The involvement of young people with an ASD diagnosis, particularly those who have received genetic testing or have participated in genomic research, would also provide the opportunity to test out the argument that genomic citizens in the making oscillate between growing genomic consumerism and limited agency (s2.4.2). This could not be explored in the present study, given that participants were not recruited because of their experiences within genomic services. It is important to note that to date there is no approved methodology to elicit the first-person perspectives of autistic people with minimal verbal abilities, for whom parents usually act as spokespeople. However, current research suggests that they are the subset of individuals on the spectrum to whom genomic information is the most relevant. Therefore, it is essential to develop methodologies to undertake research with autistic people with low IQ and communication impairments (for pioneering work in this direction see Tesfaye et al., 2019). In the future, this may

allow for a follow-up study of the present research with autistic young people, in a way that is more comprehensive of the variety of experiences on the spectrum.

Although several studies have been conducted to explore adults' attitudes towards advances in autism genomics (s3.5), their aims, objectives and methodologies differ from those of the present research. These studies have not involved adults from the general population, and many are survey-based. Most importantly, they have not adopted a method of data gathering in which participants' views are not just taken at face value, but are challenged to get to an understanding of the underpinning reasons. This is more common in empirical bioethics methodologies (Ives, 2008), particularly those that reflect the philosophical tradition of the Socratic dialogue, such as the approach upon which I have drawn in this research. Therefore, the comparisons between young people's and adults' views and attitudes that I made in s7.3.2.3 above can only be tentative. Ideally, a follow-up study conducted with adults should adopt the same aims, objectives and empirical bioethics methodology chosen in the present research and adapt them to an adult population, to allow to draw more definitive conclusions about what we can learn from the special position that genomic citizens in the making occupy in society compared to adults.

This is particularly important because some choices I made about data collection have likely probed the nuanced views reported in the result chapters. Not only was the study methodology co-developed with the NeurOx YPAG to ensure that this research would speak to young people. The tasks undertaken in Part 1 of the FGDs were in part educational, which might have also had an impact on young people's views and attitudes, even though I began them with open questions and my explanations only came at the end. However, it is important to note that the young people appeared to be quite knowledgeable about autism, except for their endorsement of some stereotypes. Thus, in many cases my educational comments only emphasized certain concepts that participants were already familiar with and had mentioned on their own (e.g. the ASD spectrum), rather than providing the young people with new information. Similarly, data analysis did not

identify any significant difference in participants' conceptualization of autism depending on the video clip shown (s4.3.4.2), which may be because no clips introduced any ideas that were not already part of participants' common knowledge of autism.

The group setting might have also encouraged participants to think of autism in a nuanced way, because exchanges of opposing ideas compelled them to reflect on and articulate the arguments undergirding their views and attitudes. This was also facilitated by the active role that I played, by challenging young people's insights with counterarguments. Additionally, the FGDs were very structured; the activities allowed participants to gradually shift from factual to normative considerations, and the investigation of both their views of autism and attitudes towards the interventions meant that by the time they were discussing ethical issues around the use of ASD genomic information they had already reflected on autism for a while. This might explain why young people made some sophisticated observations that parents who participated in past empirical studies seem to have overlooked, for instance with regard to the increased stigma that might derive from the availability of ASD treatments (s6.4.2), or the limited clinical validity and utility of PND and PGD for autism (s6.5.2).

As anticipated in s4.3.5, it should also be noted that I could not establish inter-coder reliability in this study. However, qualitative researchers have debated whether this is an appropriate and necessary tool to assess the quality of the coding frame and analysis within the goals and epistemological underpinnings of qualitative research (O'Connor and Joffe, 2020). Alternative quality criteria have been proposed, such as attention to deviant cases or thick description providing plenty of samples from raw data, which I have adopted in my research.

Coupled with my pre-existing intuitions and experiences as a researcher, on which I commented in the section about reflexivity in Chapter 4, the above considerations suggest that biases might have been introduced in the study results. Nonetheless, the present research provides evidence of methods that support young people to discuss complex ethical issues that future research aiming at

engaging children in bioethics debates should consider adopting. The only activity with which participants seemed to struggle was the one about EIs, which may be because they were new to the EI paradigm; indeed, they asked many clarifying questions. Moreover, this activity came at the end of the FGDs, when participants may have been less engaged. Thus, future research should consider a better way to elicit young people's insights on ethical issues emerging from EIs for ASD.

The philosophical analysis of the data had already allowed me to draw some considerations about the ways in which young people reasoned around the ethical issues relevant to the research topic (s7.2.2). However, because of the exploratory nature of the present study, these considerations were inevitably limited. Therefore, more systematic research (potentially including teachers and parents) should be co-produced with youth to identify age-appropriate interventions and tools to foster a generation of well-reasoned and thoughtful citizens who contribute to (the present and other) bioethics debates.

Finally, some important considerations about the interpretation of the research findings in s7.3 above are in order. First, in this study I employed a method of philosophical argumentation, and so I was only able to record the views and attitudes that the young people were conscious of and were willing to articulate in a group, rather than implicit beliefs that would be grasped by methods used in social psychology (Heine et al., 2019). Therefore, I could only advance tentative explanations about young people's (lack of) genetic deterministic and essentialist views, and hence stigmatizing attitudes. Although my suggestions corroborate recent findings in the field (Lebowitz, 2014), it would be helpful if future work could test in a more systematic way the impact that biogenetic attributions have on deterministic and essentialist beliefs in young people, and how they influence attitudes towards the use of genomic information in psychiatric settings.

Further, this thesis did not aim at providing an ethical argument to convince the reader of a certain normative claim about how autism should be conceptualized and what choices people should make about ASD genomic information. The philosophical analysis elucidated the arguments

underpinning young people's views and attitudes, and so identified a different normative framework from that of traditional biological citizens, one that takes into account the nuances of the intersection between genomics and mental health. By broadening the moral horizon of traditional biological citizenship projects, the philosophical analysis allowed me to shine a fresh perspective on Rose and Novas's account. In this sense, the approach of grounded moral analysis, which requires the iterative process of data collection and analysis to be 'repeated until the participants' experiences, attitudes, and intuitions have shaped a particular normative claim' (Dunn et al., 2012, p. 474), was drawn upon but not adopted completely in this research.

However, in my study, additional data collection would not have shaped a *particular* normative claim. On the contrary, I argue that strong normative conclusions do not fit the 'messiness' that characterizes the practical ethical context investigated in this research. By putting the empirical data under philosophical scrutiny, it emerged that uncertainty, which was a core feature of young people's contributions, resulting from their dimensional way of thinking about the research topic, added reflexivity to the discussion; in other words, it prevented participants from leaning towards clear-cut views about ASD and its aetiology, and attitudes towards the interventions, that were grounded on stigmatising and deterministic assumptions about autism, and overlooked the nuances and peculiarities of the practical ethical context at stake (s7.2.2).

Therefore, in keeping with arguments from science and technology studies (Kerr et al., 2007), this research suggests that uncertainty might be a feature that should be embraced, rather than considered as problematic in ethics discussions at the intersection between genomics and mental health. This makes it important to further investigate uncertainty not only at a conceptual level, but also as a feature that characterizes empirical decisions about complex bioethical issues, such as the ethics of psychiatric genomics. The cognitive processes that lead young people to use, and strategically bracket, uncertainty in developing and articulating moral judgements about bioethical topics should be empirically investigated. This approach would further inform efforts to develop

methods that encourage genomic citizens in the making to act as reflective citizens in bioethics debates, who integrate uncertainty as a positive aspect in their decision-making processes.

Finally, in s7.3.2.2 I argued that young people's genomic citizenship in the making should be extended beyond the case study of autism to the broader field of psychiatric genomics. Even though participants in this study oscillated between reflections on autism and other mental conditions, a legitimate concern might be that autism is to some extent exceptional among other psychiatric conditions. This is because ASD is among the few mental conditions for which tangible advances have been made in the understanding of their underlying biology; CNV testing in the context of ASD is one of the few applications of current psychiatric genomic knowledge that the International Society of Psychiatric Genetics supports (2019). Therefore, some might question whether the case study of autism genomics is generalizable to the broader field of mental health. Because the argument about extending young people's account was grounded around the categories of 'dimensionality' and 'uncertainty', which apply to any psychiatric condition, I believe that there are reasons to think that this is the case. However, future work should assess whether there might be any difference in young people's views of a range of psychiatric conditions, and how this affects their arguments about genomic interventions, and so their conception of biological citizenship.

Similarly, although participants drew a strong distinction between psychiatric and somatic disorders, future empirical and normative investigations should test the hypothesis that their account of genomic citizenship in the making could extend beyond psychiatric settings (s7.3.2.2). Such an investigation should focus on comparative questions, which in this research I only used as a methodological tool to distil the arguments underpinning young people's views and attitudes. This would allow us to evaluate whether young people's position in this study represents a form of mental health 'exceptionalism'.

7.5. Conclusion: Answering the primary research questions

By summarizing and interpreting the research findings, the present chapter sought to answer the primary questions of this research. In particular, I aimed to infer an account of the meaning that young people attribute to 'biological citizenship' in psychiatric settings; to identify the responsibilities that they promote under this account; and to explain why and how such an account should underpin and guide action about the use of genomic information in psychiatry.

I argued that children do not seem to align with Rose and Novas's ethics of active citizenship, which requires responsible citizens to draw on genomic information and make calculations in order to maximize health outcomes and minimize the risk of illness. The research findings allowed me to derive an account of young people's understanding of biological citizenship, or genomic citizenship in the making, which highlights that in psychiatric settings there is no agreed-upon distinction between health and illness, and risk calculations are usually more complicated than for Mendelian conditions. Thus, young people in this study have prioritized an ethical framework that promotes citizens' responsibility for exercising autonomous decisions, valuing and loving diversity, rejecting stigma, respecting identity and moving beyond genetic determinism and essentialism.

I argued that this is a defensible account, i.e. it should be taken seriously to drive deliberations in psychiatric genomic contexts, because it derives from young people's nuanced way of thinking of autism based on the categories of 'dimensionality' and 'uncertainty', which are mindful of the complexities arising in the context of autism genomics. Because these categories are not relevant to autism only, I suggested that the framework of genomic citizenship in the making should be applied beyond the case study of autism to the use of genomic information about other (psychiatric or non-psychiatric) conditions characterized by dimensionality and uncertainty. Finally, to answer how such an account should guide action about the use of genomic information, I compared it with

the perspectives of other autism stakeholders. I concluded that genomic citizenship in the making should not substitute other normative frameworks, but should be integrated into an expanded understanding of biological citizenship.

Chapter 8. Conclusion

This research started with a commitment to involving young people who have no 'special' relation to mental health in bioethics discussions about psychiatric genomics. Unprecedented efforts have indeed been made to advance knowledge of the biological underpinnings of mental conditions to improve their diagnosis, prediction, prevention and treatment; this will likely confront the current generation of young people with ethical questions that were less or not relevant to older generations. Thus, Chapter 1 remarked on the importance of investigating what moral concerns young people raise about the use of genomic information in psychiatric settings.

Central to discussions about advances in genomics is the concept of biological citizenship, and its related notion of genetic responsibility, as formulated by Rose and Novas. Chapter 2 described three assumptions underpinning Rose and Novas's traditional notion of biological citizenship, namely:

1. A consensus allegedly exists over where to draw a distinction between health and illness, so that the former can be maximized and the risk of the latter can be minimized;
2. A linear relationship exists between genetic risk and action, whereby genomic information is necessarily informative and actionable;
3. The neoliberal biological citizen is always in a position to make free and informed choices, and the values underpinning these align with the normative framework of biological citizenship projects.

I argued that the focus of this thesis on psychiatric genomics and the involvement of young people suggest that scrutiny should be exercised on these assumptions. When genomics is applied to mental health it becomes apparent that psychiatric conditions have an uncertain ontology, aetiology and diagnosis, and they are highly stigmatized; this causes disagreement on what constitutes 'health' and 'illness', and complicates risk calculations based on genomic information, hence

challenging assumptions 1 and 2. The peculiar position that young people as 'genomic citizens in the making' (Sabatello and Appelbaum, 2016) occupy in society suggests that children do not match the profile of the neoliberal biological citizen, hence raising questions about how young people relate to biological understandings of citizenship (assumption 3).

These considerations sparked the intellectual interest that motivated me to test the traditional notion of biological citizenship in *psychiatric settings* and from the perspective of *young people*, by addressing three primary research questions:

1. What meaning does the current generation of young people attribute to 'biological citizenship' in mental health settings?
2. What responsibilities do contemporary young genomic citizens promote under this account of biological citizenship?
3. Why and how should these responsibilities underpin and guide action about the use of genomic information in psychiatric settings?

To answer these questions, I conducted an empirical bioethics study focused on autism genomics. In Chapter 3, I explained that I chose autism as a case study not just because it has been the focus of increasing genetic and genomic interests since the 1970s, but also because ASD fits the theoretical framework developed in the background chapters due to its contested diagnosis, aetiology and nature, as well as the stigma attached to it. Moreover, prior to this research the general population, and young people in particular, had not yet been involved in the debate around advances in ASD genomics. Thus, autism was well suited for the first investigation of the moral attitudes that 'lay' young people hold towards interventions that propose to use genomic information for the prediction, prevention, and treatment of psychiatric conditions.

As Chapter 4 explained, the hypothesis that societal desire for the interventions would likely be influenced by the way in which autism is conceptualized, made it necessary to undertake a

preliminary investigation of young people's views of the traits, aetiology, and nature of autism. In this way, the empirical study provided a picture of young people's views of autism and attitudes towards the interventions, on which a normative analysis could be performed to distil the underpinning arguments, and so inform an understanding of the meaning that young people attribute to 'biological citizenship' and 'genetic responsibility' in mental health settings.

Chapter 5 and 6 showed that young people's dimensional understanding of autistic traits, severity and aetiology, and their uncertainty about the nature of autism, meant that they did not support clear-cut attitudes towards the genomic-based interventions. Because of their 'special' position in society, as younger members of the general population, they proposed an account of biological citizenship that gives space to different options about how to use ASD genomic information. This account was not based on Rose and Novas's ethics of active citizenship, but on a normative framework that values inclusivity and moves beyond genetic determinism and essentialism, hence confirming the limited portability of Rose and Novas's assumptions to the field of mental health.

By building on Sabatello and Appelbaum (2016), I called young people's account 'genomic citizenship in the making'; by moving beyond a developmental understanding of the label 'in the making', I argued that young people's account should be taken seriously because it is mindful of the complexities that arise in the field of psychiatric genomics. I advanced the hypothesis that this account applies beyond autism and psychiatric genomics, to any dimensional condition around whose nature there is no consensus. This investigation concluded that the traditional notion of biological citizenship should not be abandoned, but expanded to include the responsibilities and values promoted by young people's genomic citizenship in the making. Figure 15 illustrates the journey that the reader has undertaken while engaging with this thesis.

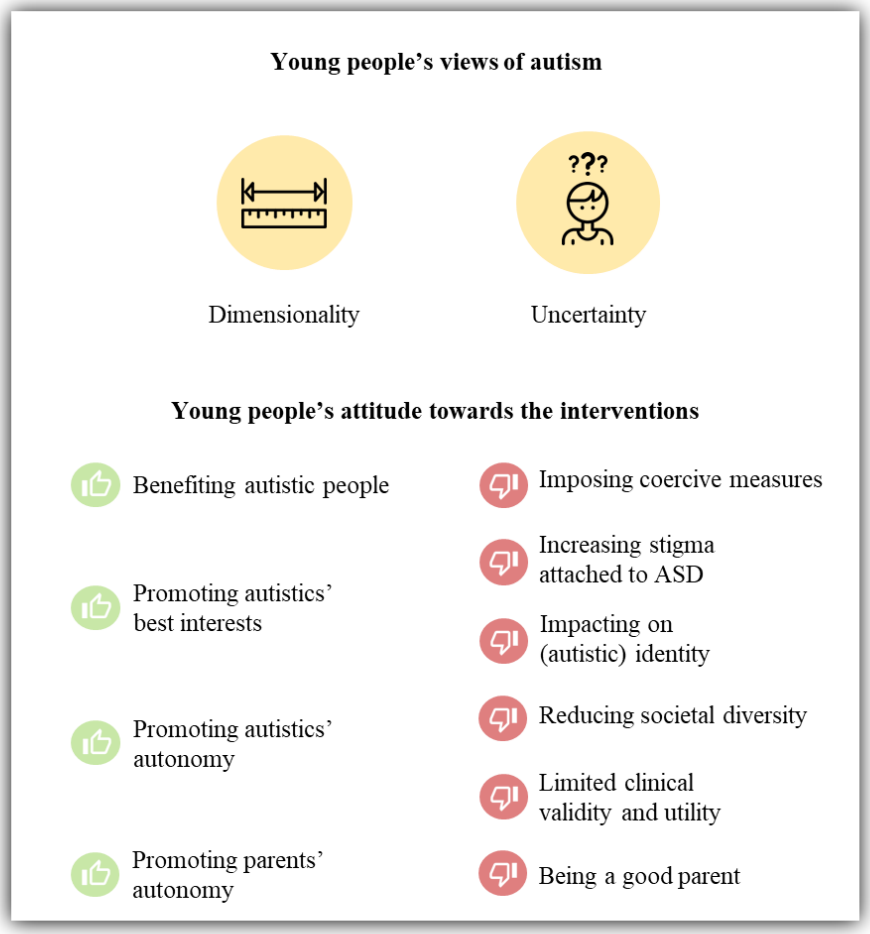
BACKGROUND

Traditional notion of biological citizenship

Need to be tested in relation to:

Mental Health | **Genomic citizens 'in the making'**

EMPIRICAL STUDY



CONCLUSION

Young people proposed a different account of biological citizenship:

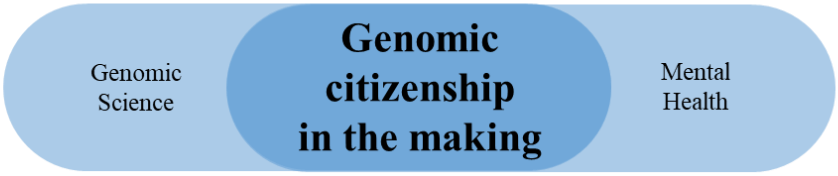


Figure 15: A graphic illustration of this thesis

The present research has made both a theoretical and a methodological contribution to the field of bioethical inquiry at the intersection of the scholarships on biological citizenship, neuropsychiatric gen-ethics and childhood participation. Although the importance of including young people in psychiatric genomic debates had already been remarked on by other scholars (Sabatello, 2017), the significance of this research is that it did so empirically, hence allowing to shed a new light on the literature by inferring an empirically grounded conception of what being a biological citizen in mental health means to young people. In this way, by using the label 'in the making' as an opportunity to reflect on what we can learn from the different role that young people play in society, I moved beyond Sabatello and Appelbaum's approach to show that young people should not be seen as deficient in biological forms of citizenship compared to adults, but as proposing a different, and yet valuable, understanding of biological citizenship.

In the background chapters, I argued that a genuine concern with children's 'special' position in society requires adults to seek their contributions in a way that accommodates the different capacities that children have compared to adults. In this research, I achieved so by working directly with young people. The consultation with the NeurOx YPAG allowed me to develop a targeted methodological design that was informed by YPAG members' experiential knowledge on how to best probe young people's insights on debates of moral and societal relevance. This has likely helped participants to develop a nuanced account of ASD, which allowed me to cast doubts on the scepticism towards young people's capacity to reflect on complex ethical issues.

By investigating and discussing how children wrestle with ethical considerations in neuropsychiatric genomics, this thesis has highlighted the considerable potential for the involvement of young people in bioethics research and debates. Future work should engage with the limitations highlighted in Chapter 7 to create new opportunities for considering what bioethicists can learn from listening to young people's views and attitudes towards currently pressing debates.

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Appendices

Appendix A: Overview of socio-empirical papers on autism genomics

Author and year	Focus	Population	Objective	Method	Main findings
Chen et al., 2015	Prenatal genetic testing (PGT) for ASD	42 parents of autistic children, USA (Texas)	Investigating parents' attitudes towards prenatal genetic testing for ASD and termination of pregnancy	Interviews	*Attitudes towards PGT* Over half participants willing to undergo PGT for autism Arguments in favour of PGT for autism: better preparation for birth of a child with ASD, early and better medical treatment, termination of affected pregnancy, contribution to research, and curiosity Arguments against PGT for autism: irrelevance of test result for reproductive decisions, religious beliefs, potential PGT-related anxiety during pregnancy, apprehensions about test limitations Uncertain attitudes toward PGT for autism: depending on condition of pregnancy, stage of gestation, cost, test procedure, and accuracy *Attitudes towards termination of pregnancy* Arguments in favour of continuing affected pregnancy: preparation for birth of a child with ASD, bond with or acceptance of existing ASD-affected children, apprehensions about test limitations, religious beliefs Arguments in favour of terminating affected pregnancy (primarily Asian parents): stigma around ASD in Asian community, ASD as burden to affected child, family, and society
Reiff et al., 2015	Clinical genetic testing	57 parents of autistic children who	Investigating parents' experiences	Semi-structured telephone	Most parents reported that CMA was helpful for their child and family. *Survey*

	(CGT) for ASD	remembered that their child had had CMA, USA. Of them 50 also completed survey	with CGT for their children and perceptions about the usefulness of test result for child and family	interview and survey	60.8% parents reported that the test result has been moderately, very, or extremely helpful both for the tested child and their family. Result extremely helpful only for parents receiving pathogenic result. *Interviews* Factors increasing usefulness: medical care (e.g. ruling out a genetic condition; information about kidney problems, bad reaction to certain anaesthetics, susceptibility to cancer, heart diseases, etc.), educational and behavioural interventions, causal explanation, information for family members (better understanding of recurrence risk, but reproductive decisions not necessarily affected; networking with family members with same variant or with organizations; influence on financial planning; removing guilt), advancing knowledge (hope for potential treatment or pharmaceutical both for own child and other families; potential diagnostic benefits from increased understanding of genetic of ASD). Factors limiting utility, increasing uncertainties and negative outcomes: negative or VUS results did not lead to any specific changes for child or family; lack of new treatment, or no change in family day-to-day life; interventions already being received based on ASD diagnosis and not affected by genetic test result; only confirmation of what family already knew or suspected about the child; personality of the child more informative than genetic test result; psychosocial harm from positive test result (e.g. blaming one side of the family, contribution to sense of guilt); confusion or misunderstanding of genetic information
Xu et al., 2016	Clinical genetic testing for ASD	45 parents of autistic children, USA (North Eastern region	Investigating parents' awareness and perceptions of	Interviews	*Awareness* None of the parents had ever heard of the test before *Perceptions*

		and North Carolina)	CMA, and motivating factors and barriers to test		Majority of parents postulated positive attitudes towards test *Motivating factors* Identification of potential cause of child's ASD (including peace of mind to parents and relieving them from guilt), better preparation for another autistic child, informing future reproductive decisions (including through knowledge about recurrence risk), early intervention opportunity for autistic child, research to benefit other families with autistic children, benefits to autistic child (therapies, medical treatments), confirmation of ASD diagnosis *Barriers* Cost of CMA, risk/pain of CMA, fear of test results, little to no motivation to test, transportation and/or scheduling issues, lack of knowledge and understanding of CMA
Li et al., 2016	Clinical genetic testing (CGT) for ASD	42 parents with at least one autistic child, USA (Texas)	Investigating parents' information needs regarding ASD genetic testing	Semi-structured, in-depth interviews	Majority of parents (83%) had never received information about CGT from their doctors Most parents (86%) interested in having more information. Main topics of interest: costs/health insurance, benefits (value of confirmatory diagnosis, other potential benefits for affected families and ASD community in general), accuracy, test procedure, potential physical harm, confidentiality, previous utilization by other affected families, eligibility criteria Those not interested provided the following reasons: testing would not improve current situation, testing is not among family's priorities
Hens et al., 2016b	Clinical genetic testing for ASD	15 Belgian professionals (clinical genetics, psychiatry, neurology,	Autism healthcare professionals' perspectives of genetic counselling,	Semi-structured interviews	Participants considered genetic counselling and testing useful after ASD diagnosis, but benefits for diagnosed children only in a subset, especially if known variant with known action plan and prognosis Other benefits for parents: explanation for child's autism, and reproductive information.

		psychology, or educational science) with ASD as primary expertise	testing, and reproductive choices		Risks for parents: inducing guilt and blame Use of PGD and PND acceptable only if additional factors could decrease quality of life or if too high burden of having an (extra) autistic child for parents
Johannessen et al., 2017	Clinical genetic testing (CGT) for ASD	1455 parents of autistic children from Norwegian Autism Society, Norway	Investigating parents' attitudes towards ASD genetic testing	Questionnaire	Most parents had positive attitudes towards CGT. Arguments in favour: 76% would undergo testing provided it could give causal explanation to their children's ASD, 74% would undergo testing provided it improved opportunities for EI Arguments against: 49-67% agreed that testing could negatively impact health insurance, increase anxiety for child's future, cause family conflict; some parents were against testing because of less optimism regarding positive effects Parents prefer that CGT is offered to those with autistic child (65%), when the child's development deviates from normal (48%), or before pregnancy (36%) Prenatal tests were not the preferred option
Narcisa et al., 2012	Genetic testing to assess risk for ASD	162 parents of at least one autistic children, USA	Investigating parental experiences of ASD diagnostic process, anxiety levels about ASD recurrence risk, and opinions about the use of genetic testing	Survey	*Diagnostic process* Mean time to ASD diagnosis and age at diagnosis after parents first express concerns: 35.2 months and 56.6 months; 72% parents felt that there was a delay in diagnosis Majority of participants believed genetic factors contributed to the development of ASD; 82.1% indicated that it was a combination of genetic and environmental factors; 11.9% indicated that ASD was almost entirely a result of genetic factors; 6% indicated they believed ASD was caused almost entirely by non-genetic factors More than 2/3 children did not receive testing; minority of parents were unsure whether their children had had testing *Anxiety levels*

			to assess risk for ASD		52% parents whose families included a younger undiagnosed child (< 48 months) felt the younger child was somewhat/highly likely to develop ASD; 48% indicated they had a somewhat or very high level of anxiety associated with the possibility that the younger undiagnosed child could develop ASD *Genetic risk assessment test* 80% parents would want to pursue genetic testing that could identify risk in younger siblings even if it could not confirm or rule out the diagnosis. Reasons in favour of testing: Earlier evaluation and intervention, closer monitoring, and lessening anxiety Reasons against testing: risk of increasing anxiety and labelling child, lack of awareness of the utility of test result
Xu and Floyd, 2017	Autism genomic research and clinical genetic testing for ASD	204 parents of autistic children, USA (Easter North Carolina)	Examining the association between parental perceptions regarding autism research and their intention to take their children to undergo recommended genomic tests (CMA)	Online survey	High parental intention to test their children (84.3%), despite evidence of low CMA uptake Parents desiring to help and with favourable intentions towards ASD genetic research were more likely to test Parental intention to test associated with the prospect of contributing to research. Intention increases with belief that genetic testing advances ASD research. In turn, this may enhance autistic child's prospect
Singh, 2014	Autism genomic research	23 sets of parents who donated their	Investigating different narratives of	In-depth interviews	Three narratives of participation: 1. Altruistic parent: Form of participation that is reflective of traditional biological citizenship that creates forms of

		family's blood and medical information to an autism genetic database (The Simons Simplex Collection), USA	parents' participation in genomic research within the context of biological citizenship		biosociality based on shared corporeal vulnerability, somatic suffering, and genetic susceptibility 2. Obligated parent: Helping children in any possible way; based on need for knowledge that can potentially help children, but not in terms of finding a 'cure' for autism 3. Diagnostic parent: Participation based on the opportunity to receive a free diagnosis as part of the study. Range of social circumstances surrounding participation took a prevalent role over concerns about traditional bioethical principles (e.g. privacy or confidentiality). Alternative form of contextual bioethics and moral reasoning to participate based on the structural limits and social complexities of their situations: parental responsibilities, hopes in genomic science, and need for early diagnosis and access to services. Narratives make explicit the moral reasoning and personal negotiations of risks and benefits of participation that move beyond bioethical boundaries based on specific principles and their applications. This socially complex and situated nature of participation leads to a new form of biological citizenship, in which structural constraints matter and should be taken into account.
Johannessen et al., 2016	Autism genomic research	1455 parents of autistic children, Norway	Investigating parents' attitudes, fears and expectations toward autism genomic research and procedures in autism	Questionnaire	Motivating factors for participation in research: gaining more knowledge of the causes and mechanisms of autism (83.6%) and contributing to the development of better treatments in the future (63.7%) Positive attitudes towards storing genetic information (54.3%); importance of confidentiality of data (82.9%), and information about the purpose (89%) and progress (83.7%) of research

			genomic research		
Hens et al., 2016c	Autism genomic research	15 Belgian professionals (psychologists, clinical geneticists, educational specialists, paediatricians, child neurologists, child psychiatrists) with ASD as primary expertise, including people with experiences in caring for children with severe mental retardation	Mapping ethical issues experienced by professionals involved in genetic research on autism	Semi-structured interviews	The heterogeneity of autism affects research ethics in the following ways: 1. Selection of research participants 2. Aim of research 3. Research procedure (e.g. modified consent form, or blood draws) Potential benefits from research: improved diagnosis through biomarkers; better understanding of link between genetic variants and specific developments in central nervous system; better understanding of the development of autism, and so more accurate prediction of development of children with specific genetic subtype; better prognosis; earlier detection of autism; better pharmaceuticals; prevention of autism through prenatal screening and information about reproductive choices Concerns about implications of research: psychological impact of knowing too early about ASD susceptibility; doubts about desirability of medications as effort to normalize autistic people, and of prenatal screening; scepticism around extent of research contribution; uncertainty around research contribution to autistic people's quality of life Uncertainty around aim of research pervaded by disagreement around the conceptualization of autism
Tabor et al., 2011	Autism genomic research and returning results	23 parents of children enrolled in genetic research about autism or diabetes, USA	Exploring how parents make decisions about enrolling their children in research and their perspectives	Phone interviews	*Participation in research* Factors influencing parents' perspectives on children's participation in genetic research: protecting child from unnecessary harms, time and convenience, opportunity for altruism and positive action, 'rewards' of research participation (e.g. better interventions and services for child), relevance of the study to the family, and perceived importance to society

			about receiving children's research results		*Return of results* Several implications of hypothetical scenarios about return of results: psychological impact of results on parents themselves (blame, guilt, fault, anxiety, stress; relief from guilt, empowerment from increased knowledge), clinical utility (about which parents of autistic kids spoke more vaguely), reproductive utility (which was discussed primarily by parents of autistic kids), personal utility, and results as an incentive for further research participation
Trottier et al., 2013	Autism genomic research and returning results	9 parents of autistic children whose family enrolled in genetic research, Canada (Toronto)	Investigating parents' motivations to participate in autism genetic research, the perceived values in participating and having an actual or hypothetical genetic research result, and attitudes about the certainty of a genetic research result	Semi-structured interviews	*Motivations to participate* Combinations of benefits to the family and others. Altruism had many forms, including incidental advantage, contributing to advancing the field for the benefit of the autism community including themselves; some felt obliged to enrol if personal or community gains expected *Expectations about genetic results* All respondents would want a research result if it were available, but benefit even without results (e.g. feelings of being proactive and taking control over uncertain situation, connecting with ASD experts, and other families, hope for the future) Value of having genetic result: expected or actual feeling of relief at having a tangible genetic explanation for the cause of child's autism; means for personalizing interventions, for family planning, and for predicting other potential health concerns; 'proof' that there is a medical condition, and so potential increase in state supports and resources, and increased awareness in society that could lead to acceptance of autism as a biological rather than psychiatric condition, diminished negative judgement from the general public, including stereotypical views about autism special talents

					None of the respondents spontaneously mentioned ambiguity or uncertainty with regard to a hypothetical genetic research result
Miller et al., 2010b	Returning autism genomic research results	60 parents of autistic children from Canada; 23 researchers from Canada, US, and Europe	Investigating researchers' and parents' experiences and expectations regarding the disclosure of genetic research results, particularly VUS	Focus groups and interviews with parents; interviews with researchers	Mediating factors for result disclosure: personal utility or perceived meaning for participants, including non-clinical benefits (reproductive risk, causal explanation, bringing 'peace of mind' or reduced sense of blame); validity of test results (mentioned mainly by researchers); participants' commitment to certain ASD aetiology (genetic v. social factors) Concerns about result disclosure: scepticism about non-medical benefits in the long term; risk of blaming one side of the family; different meanings of validity of test results by discipline (biological and clinical norms support return of individual results; statistical norm consider results meaningful in the aggregate only)
Baret and Godard, 2011	Returning autism genomic research results	158 parents of autistic children participating in genetic research, Canada (Montreal)	Parents' perspectives on returning results of autism genetic research	Questionnaire	97% strong desire to receive results, either general or individual. Two profiles of people who want to receive individual findings: people concerned with the possible transmission of autism (20%), who would make practical use of research results (preparing for the future, making reproductive decisions); people who want to know more (15%) with no associated intentions other than advancing scientific knowledge, bringing relief or understanding Participants did not perceive any harms in receiving individual results, even if listed in questionnaire Participants understood distinction between clinical and research settings and did not have clinical expectations
Hayeems et al., 2011	Returning autism genomic	343 cystic fibrosis (175) and autism	Exploring factors influencing researchers'	International, cross-sectional survey,	Most participants (80%) agreed in principle that clinically significant results should be reported to participants, but also that provisional scientific information is potentially harmful for research participants

	research results	genetics (168) researchers	judgements about reporting genetic research findings to individual study participants	quasi-experimental design	Diversity of judgements about meaning of 'clinically significant' results that should warrant reporting. Moderating factors: 1. Scientific: replication, robustness, intentionality (non-incidental), and disease context. ASD findings engendered 35% less confidence than CF findings; researchers were half as likely to support reporting of ASD findings compared to CF findings 2. Capacity of research team to explain result: 40% less likely to support reporting if team lacks capacity to explain results and provide medical advice 3. Type of research ethics guidance: 40% less likely to agree that result should be reported if consent form not specific about how to manage result reporting 4. Researchers' disease community (CF or ASD): ASD researchers 40% less likely than CF researchers to be confident in a given finding; ASD researcher 40% less likely to support result reporting 5. Researchers' primary role (clinical, molecular, statistical): clinical interpretative role two times more likely to be confident in the clinical significance of a finding and 1.5 more likely to support result reporting; statistical role 30% less likely to support reporting 6. Beliefs about general reporting obligation: endorsing belief meant 2.8 times more likely to feel confident about clinical significance and four times more likely to support reporting
Pellicano et al., 2014	Priority-setting in autism research	*Focus groups/interviews* 14 autistic people, 27 parents of autistic children	Investigating autism community's views of current pattern of UK autism research, and	Interviews, focus groups, online survey	*Focus groups/interviews* Unanimous agreement that current UK autism research focuses on biomedicine; exception was parents, who believed that current research focuses on behaviour, environmental impact on autism, education Unanimous agreement that most research focuses on children; for some parents focus also on high-functioning autistics

		from those with limited spoken language to high functioning, 20 practitioners (speech/language therapist, teachers, educational psychologists), 11 autism researchers, from UK *Survey* 122 self-identified as autistics; 825 parents/carers; 24 sons, daughters, siblings, 426 practitioners (e.g. teachers, speech/language therapists, support workers, psychologists, paediatricians), 120 ASD researchers, UK	differences among stakeholders		Unanimous disappointment with current emphasis on biomedical research, but researcher to lesser extent; for many participants research should be more evenly distributed among primary research areas For future research, all stakeholders, particularly autistics, parents, and practitioners prioritized research into issues of immediate practical concern, particularly services and supports, knowledge about autism, and research logistics; but some differences: autistic adults did not support interventions meant to normalize people (core symptoms) *Online survey* All stakeholders dissatisfied with current pattern of UK research priorities Convergence between autistic adults, family members, practitioners and researchers on two top research areas: improving the life skills of autistic people, identifying how public services can best meet the needs of autistic people Open questions about the topic of research that stakeholders would like to be researched in the coming decade: convergence towards research that would make a difference to their everyday life
Frazier et al., 2018	Priority-setting in autism research	485 autistic adults, 4400 family members, 401	Investigating stakeholders' value and importance of	Online survey	Preference for applied (including broad areas of screening/identification tracking and interventions) relative to basic science (including discovery of causes and

		researchers, 1306 clinicians/educators, 670 others, from US, Canada, UK, Europe, Australia/New Zealand, Asia, South America, Middle East	broad lines of scientific inquiry and specific content areas relevant to autism science funding		understanding of ASD biology) topics, but both important; people with autism tended to rate less favourably Highest rating on research focused on co-occurring conditions, health and wellbeing, adult transition, and lifespan issues; no significant difference across different groups; correlation between increased age and importance of adult transition and lifespan issues Open ended questions about areas to support or avoid funding: studies of genetic markers and prenatal screening as areas to avoid; small subset of respondents, most with autism, expressed disapproval of all autism-related research or approval restricted to research focused on co-occurring condition, adult transition, lifespan issues, health and wellbeing; more than half did not identify research areas to avoid; strong desire to include more people with autism and their family members in research funding process
Russell et al., 2017	Pharmaceutical interventions for autism	Autistic adults, parents of autistic children, autism professionals (teachers, clinicians, researchers, nurses), [demographic information not available]	Scrutinizing patient and public involvement by a pan-European biomedical consortium working to develop drugs to treat autism	Open comments on promotional video by consortium project agenda	Two major themes: 1. Autism is not a suitable target for treatment: uneasiness with description of autism as 'disease' or as having biological cause; curing equated to changing identity; scepticism about single treatment for heterogeneous condition; narrative of neurodiversity movement; impossibility of treating traits in isolation; risk of eliminating valuable traits but also acknowledgement of difficulties that require support 2. Circumstances when drug treatment is acceptable: short term gains (managing stressful situations or targeting co-occurring problems); importance of autistic person's agency (being in control); against autism as a generic target; concerns about 'who should decide?', about use of psychotropics as a convenient form of social control

					Different view from parents of severely autistic children: agreement with message that autism is a disease that can be harmful; support for drugs development that could be used in combination with therapies Call for greater involvement of autism community in future biomedical research, and criticism to Autism Speaks
Fletcher-Watson et al., 2017	Early autism research	2317 autistic adults, parents of autistic children, practitioners in health and educational settings from Czech Republic, Finland, France, Israel, Italy, Macedonia, Norway, Poland, Portugal, Spain, UK	Investigating autism community's opinions about early autism research to provide a foundation for early autism researchers wishing to engage with stakeholder groups	Focus group pilot, online survey	*Focus groups* Main themes related to early autism research: use of 'at-risk' language; intervention component in early autism research; transparency between research participants and researchers *Survey* Participants' positively disposed towards early autism research, but autistic adults' overall less favourable attitudes; overlap among groups regarding priorities for the field and preferred language to describe infant research participants Participants ranked genetic basis of ASD and early ASD signs in infants as the most important questions scientists should be asking about early autism Participants ranked early identification, better clinical knowledge of early signs, and for some stakeholders provision of help to develop skills that prevent children from later receiving an autism diagnosis as top goals early autism research should focus on; for autistic adults environmental factors and unique development as significantly more important goals, while early identification as less of a priority Preferred language to refer to infant research participants: dislike of 'at risk', except for healthcare practitioners; 'infants with high autism likelihood', 'infants with higher chance of developing autism' as preferred terms
Fletcher-Watson et al., 2019	Early autism research	1040 parents of autistic children, from UK, Czech	Investigating factors influencing	Online survey	*Attitudes* Large majority of parents showed positive attitudes, associated with greater access to services, higher service

		Republic, France, Finland, Italy, Israel, Norway, Portugal, Spain, Macedonia, Poland	parents' attitudes towards early autism research; parents' expectations about availability of intervention, acceptable forms of measurement, preferred mode of contact, sharing information		quality ratings, and higher rates of intellectual disability among their children; 87.5% parents responded 'Yes, definitely' when asked whether they thought that autism research should be done *Expectations* Parents valued scientific goal of the research academic status of research, but for half intervention component would be essential requirement for participation; presence of intervention component was ranked as less important in decision-making about participation relative to more overarching factors (e.g. what study involves for family). Positive attitudes towards all measures, but less towards brain scans for children Parents valued direct contact with research team and openness in data sharing, particularly full disclosure of all child assessments (84.9%)

Appendix B: Invitation Emails

The Invitation Emails originally presented here contain information that cannot be made freely available via ORA.

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Appendix C: Information Sheets

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Appendix D: Examples of advertising materials





Appendix E: Ethics approval letters from the University of Oxford CUREC

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Appendix F: Assent and Consent Forms

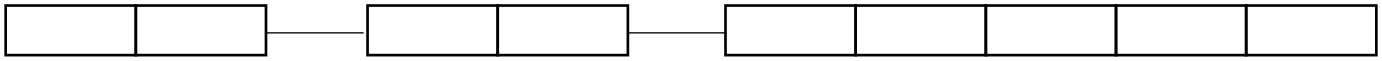
The Assent and Consent Forms originally presented here contain information that cannot be made freely available via ORA.

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Appendix G: Demographic questionnaire

Venue ID Code: Year: Focus group identification number:



Young people's moral attitudes towards autism
Young Person Demographic Questionnaire

Please fill in the following

How old are you?	_____
Postcode	_____
What is your gender?	Male ☐ Female ☐ Other ☐ Prefer not to say ☐
What is your ethnicity?	White British ☐ White Irish ☐ Other White Background ☐ Mixed White and Black Caribbean ☐ Mixed White and Black African ☐ Mixed White and Asian ☐ Other Mixed Background ☐ Indian ☐ Pakistani ☐ Bangladeshi ☐ Other Asian Background ☐ Caribbean ☐ African ☐ Other Black Background ☐ Chinese ☐ Other ethnic group (please state) ☐ _____

	Prefer not to say ☐
What is your first language?	English ☐ Other (please specify) ☐ _____
What is your favourite subject in school?	English ☐ Mathematics ☐ Science ☐ Art and design ☐ History ☐ Languages ☐ Geography ☐ PSHE ☐ Other (please specify) ☐ _____
What are your current living arrangements?	Parental home ☐ Mother's home ☐ Father's home ☐ Foster carer's home ☐ Shared accommodation (not with family) ☐ Other (please specify) ☐ _____
What is the marital status of your biological parents?	Married and cohabiting ☐ Cohabiting ☐ Separated ☐ Divorced ☐ Other (please specify) ☐ _____
What is your mother/legal guardian's occupation?	If applicable: _____ Full Time ☐ Part Time ☐ Unemployed ☐

	I do not know ☐
What is the highest level of education your mother/legal guardian has received to date? If currently enrolled, mark the previous grade or highest degree received:	Primary Education ☐ Some Secondary Education ☐ GCSE ☐ Some College ☐ A-levels ☐ Undergraduate Degree (BA, BSc etc.) ☐ Postgraduate Degree (MA, MSC etc.) ☐ Doctorate (PhD) ☐ Other (please specify) ☐ _____ I do not know ☐
What is your father/legal guardian's occupation?	If applicable: _____ Full Time ☐ Part Time ☐ Unemployed ☐ I do not know ☐
What is the highest level of education your father/legal guardian has received to date? If currently enrolled, mark the previous grade or highest degree received:	Primary Education ☐ Some Secondary Education ☐ GCSE ☐ Some College ☐ A-levels ☐ Undergraduate Degree (BA, BSc etc.) ☐ Postgraduate Degree (MA, MSC etc.) ☐ Doctorate (PhD) ☐ Other (please specify) ☐ _____ I do not know ☐
Have you ever heard of autism or Asperger syndrome?	Yes ☐ No ☐ Prefer not to say ☐
Have you been clinically diagnosed with autism or Asperger syndrome?	Yes ☐ No ☐

	Prefer not to say ☐
Do you know anyone who has autism or Asperger syndrome?	Yes ☐ Please specify your relationship with them: _____ No ☐
How often do you usually see the people you know with autism or Asperger syndrome?	Once or twice a year ☐ Once or twice a month ☐ Once or twice a week ☐ Every day ☐ Not applicable ☐

Appendix H: Topic guides

H.1 Topic guide Part 1

Secondary questions about young people's views of autism:

- What do young people understand autism is?
- What do young people think autism is caused by?
- Do young people consider autism as a disorder, a disability, or a difference (and why)?

Introduction

Hi, my name is Arianna. I am a researcher at the University of Oxford. Thank you for agreeing to participate in this group discussion. Would you mind introducing yourselves to everyone in the group? / Do you already know each other?

My team and I are trying to explore how young people, like yourself, understand autism and its causes, and how genetic information about autism should be used in medical research and clinics. We want to understand things from your perspective, so YOU are the expert here, not me. I'm learning from you. If you want to stop at any point or ask a question, let me know, ok?

Because I don't want to forget what you are telling me, is it ok if I record this discussion? Nothing you say will have your name on it and this recording will not be shared with others like your family, it is private information. If this sounds ok, **please fill out your assent/consent forms, as well as the short demographic questionnaire.**

To give you an overview of what we will be doing today, the study has two parts. In the first hour we will have a discussion on what you think autism is; this discussion will include some group activities that I will ask you to do. We'll then take a small break. After the break, we will have another discussion (about 1 hour) to discuss whether and how you think that we should use information for the prevention and cure of autism in medical research and in the clinics. Again, the discussion will involve some fun and interactive activities.

Activity 1: Introductory questions on autism spectrum disorder

What it is, familiarity with autism, autism as a spectrum disorder

- *Brainstorming activity on autism:* Can I start off by asking what you know about autism? *The researcher will note down what participants say on a piece of paper*

Prompts: Can you tell me what comes to your mind when you hear the word 'autism'; can you think of any difficulties that people with autism experience; can you think of any strengths that people with autism have; have you ever heard of Asperger syndrome (AS); do you know what it is. *If someone in the group says that they have autism/AS:* How would you explain to the group what autism/AS is. *Participants will be told that from this point on 'autism' will be used to refer to AS too.*

- Do you personally know anyone who has autism?

  Prompts: How can you tell that they have autism; what sort of behaviours do they have (give examples); do you see any of their behaviours as difficulties/strengths (give examples). *If someone says they have autism:* Can you describe any difficulties that you think you may experience because you have autism; can you describe any strengths that you think you have thanks to autism. *If no one personally knows somebody who has autism:* Do you know any autistic characters from books/movies/TV series? If yes, can you describe them? If not, where have you first heard of autism?

- Please comment on this animated video on autism.

  If the group has mainly discussed difficulties https://www.youtube.com/watch?v=7JdCY-cdgkI

  If the group has mainly discussed strengths http://www.healthtalk.org/peoples-experiences/autism/life-autism-spectrum/making-friends-social-life-autism (social communication difficulties)

  or https://www.ambitiousaboutautism.org.uk/hidden/my-sensory-world-by-louis-morel (hypersensitivity)

  or http://www.healthtalk.org/peoples-experiences/autism/life-autism-spectrum/obsessions-autism (restricted, repetitive interests)

  Prompts:

  - Would you like to change/add anything about/to what you said earlier about autism; did you learn anything from this video that you did not know about autism; is there anything about this video that you do not agree with or that you want to criticize and, if so, why. *Depending on participants' responses they may be asked to comment on specific bits of this video to learn about and reflect on two main autistic traits (social communication difficulties and restricted and repetitive behaviours).*

- *If someone in the group says that they have autism:* Do you recognize yourself in anything that was said in this video; is there anything about this video that you do not agree with and, if so, why.

- *Brainstorming activity on the spectrum nature of ASD:* Once I read this quote by an autistic man: 'If you have met one person with autism, you have met one person with autism' (Dr Stephen Shore). What do you think Stephen is trying to say with this sentence?
  - Autism is also called 'autism spectrum disorder'. Do you know why? Prompts: What do you think the word 'spectrum' means here? How do you see that in your friends who have autism?
  - The video explains that some autistic people are good at tasks that other people find difficult, such as mathematics, drawing, music etc. At the same time, there is evidence that one-third of autistic people are minimally verbal. What do you think this tells you about autistic traits (skills, capacities, difficulties)?

Activity 2: Understanding of autism aetiology

Genetic and environmental causes, multifactorial condition

- *Brainstorming activity on the risk factors for autism:* Why do you think some people have autism? Can you tell the group two or three factors that you think contribute to autism?
  - *If someone says that they have autism or that they know a person who has autism:* do you know when you/your relative/your friend/etc. started to show symptoms of autism; how do you think that worked; do you know if anyone else in your/their families has autism and, if so, why do you think this is.
- *The researcher will have a rounded magnet (no words on it), and a whiteboard with the letter E (environment) and the letter G (genes) written on two opposite sides of the whiteboard; a black line will link the two letters.* Please place this magnet on the line to say whether you think autism is mostly determined by genetics or by the environment. *Ask them to justify their choice.* Prompt: Do you think that an 'autistic gene' exists or do you think that a group of genes/genetic changes contribute to the development of autism?
- *Participants will be given one set of magnets, with the following items written on: genes, having older parents, vaccines, exposure to high level of testosterone (a male hormone) in the womb, complications during pregnancy/birth, diet, cold mothers, bad parenting, having older siblings with autism. They will then be told:* On these magnets you can read factors that may or may not increase the risk of developing autism. Please, place the magnets on the board

under Y (yes), N (no), or ? (maybe), depending on whether you think that they may contribute to autism or not.

  - Do you think that any of these factors alone may cause autism? If so, which one and why?
  - Among the group of factors that contribute to autism, which ones do you think are most likely and least likely to be risk factors for autism? Why?

- Please comment on this animated video on the risk factors for autism (1'52"' - 2'50") https://www.youtube.com/watch?v=ouQHa-o5J-w *This video will only be shown if participants present very wrong ideas about autism (e.g. autism is caused by one single gene in all cases)* Prompts:
  - Would you like to change/add anything about/to what you said earlier about the risk factors for autism; did you learn anything that you did not know about the risk factors for autism from this video; is there anything about this video that you do not agree with and, if so, why. *The researcher will go back to the line on the whiteboard and will ask participants whether they would like to move the magnet more towards either E or G, or if they want to keep it on the same point*
  - Prompts: After watching this video, do you think that autism is most likely caused by one single (genetic or environmental) factor or by a combination of different factors? Can you think of any other conditions to which this applies?
  - *Depending on participants' answer to this last question, they may be shown a few seconds (7'00'' - 8'02'') from the following video which explains that in some cases autism is part of the phenotype of a one-single-gene disorder, while other cases are caused by a combination of genes and developmental processes: https://www.youtube.com/watch?v=wKlMcLTqRLs*

Activity 3: Understanding of neurodiversity

Autism as a disease, a disability, or a difference; Neurodiversity; Autism vs disabilities

- *The researcher will write S (strengths) and D (difficulties) on the whiteboard and underneath these letters they will write the ASD strengths and difficulties that participants have mentioned in Activity 1. If needed, the researcher will ask them to think of more S and D. A spiky profile may be used here to visualize the fact that people with autism do not have a typical average ability across different tasks, but they have capacities that are at the*

extremes. Would you say that someone who has all these traits has a disease/disorder, a disability, or just a different personality? Why? Where do you see the disorder emerging?

- Now let's focus on the difficulties. Do you think a person who has these difficulties should try to get rid of them at all? If yes, how should they change them? Prompts: Should they change themselves to be able to work well in society?
- Do you see any other way in which these difficulties could become non-difficulties? Prompts: What can their families, friends, societies do? Should their families, friends, societies do something?

- Some deaf people argue that although they have a physical impairment (they cannot hear), it is society that makes them disabled, for instance by taking little or no care of the fact that they need visual aids to use certain services. They argue that there is nothing wrong with them, they are just different – some of them are proud of belonging to the Deaf culture - and that societal barriers should be removed so that they can enjoy the same opportunities as those who can hear. Do you think the same applies to autism i.e. it is society that is not allowing them to function, rather than their impairment? Or do you see a difference between the argument put forward by people with physical impairments (e.g. hearing loss, inability to walk, and short height) and autism, which affects people's mind and behaviour?
- *Brainstorming activity on neurodiversity:* Have you ever heard the word 'neurodiversity'? Can you tell me what comes to your mind when you hear the word 'neurodiversity'? What are the implications of this concept on the way we should treat people with autism? *Depending on participants' responses, they may be asked to read a leaflet on neurodiversity. They will then be asked the following questions:* Do you agree or do you disagree with what the neurodiversity movement says? Why?

Recap of the discussion

At the end of FGD_1 the researcher will clearly reiterate what is and is not known about the various categories discussed within the group, in order to make sure that participants do not walk away from the study with incorrect ideas. As we have reached the end of this session, it is important to summarize what we have learned about autism.

- Autism spectrum disorder (ASD) is characterized by social communication difficulties and restricted and repetitive behaviours. ASD also provides people with strengths, such as paying particular attention to details and holding different world-views.

- Autism exists on a spectrum of symptoms, which means that even if two people have the same diagnosis of autism, they may present very different traits.
- Scientists are still searching for specific mechanisms that may cause autism. However, they know that 'the autistic gene' does not exist, as most ASD cases are the result of a combination of genetic and environmental factors. At the moment we do not have enough knowledge to link specific genes to certain degrees of severity of autism.
- The neurodiversity movement argues that autism is a valuable part of the variety of human traits and that there is no right or wrong way to develop, which implies that we should not try to cure or prevent autism.

Concluding questions

- Is there anything else that you would like to say?
- Is there anything that we have not covered?
- Thank you very much for your time.

H.2 Topic guide Part 2

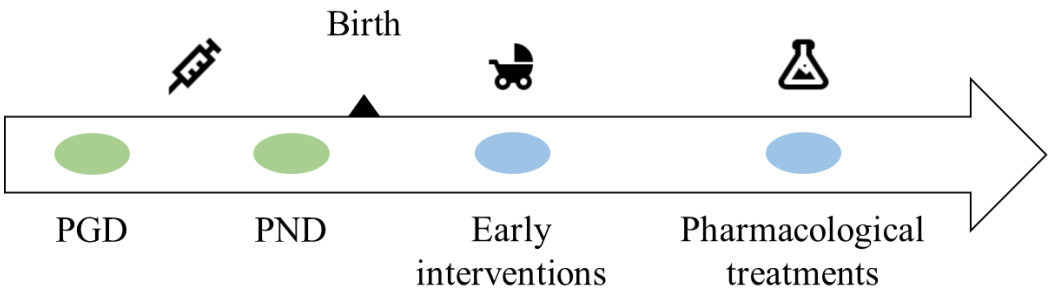
Secondary questions about young people's attitudes towards the interventions to predict, prevent, and treat autism:

- Should we welcome genomic research to develop pharmacological treatments for autism? Why?
- Should parents be allowed to use genomic information about their embryos' and foetuses' predisposition to autism to make reproductive choices? Why?
- Should we test babies for their genetic predisposition to ASD for early intervention purposes? Should we enrol high-risk children in early intervention programmes? Why?

Introduction

At the moment we are still uncertain about how autism develops. However, scientists have recently put much effort in conducting genetic research on autism. This is because they hope to achieve a better understanding of the biological mechanisms that contribute to the development of autism in

order to be able to intervene at different stages of life to prevent, cure, or reduce the impact of autism. As you can see from this figure, genetic information about autism can be used before birth and after birth. Before birth it can be used to select embryos with a decreased risk to develop autism, or to terminate foetuses that are at increased risk of autism. After birth it could be used to monitor a child known to be at high risk of autism, by intervening early to slow or stop the progress of autism if the child started to show any symptoms; or to develop pharmacological treatments to cure autism. In the next hour or so we will discuss the ethical issues that emerge from using genetic information to intervene at these different stages. *When discussing the different interventions, the researcher will remind participants where the discussion is at along the arrow.*



Activity 1: Attitudes towards curing autism
Genetic reductionism and essentialism

Let's start with a role play game to discuss a potential treatment or a cure for autism. Imagine that, thanks to genetic research, scientists have acquired so much knowledge about the mechanisms that contribute to autism that they claim they have developed a pill to cure autism. *Participants will be told that a cure for autism does not actually exist yet*. Can two volunteers read the following dialogue? This dialogue is based on the opinions expressed by two autistic young people, Sue Rubin and Naoki Higashida, on the potential development of a cure for autism *(please refer to http://edition.cnn.com/CNN/Programs/presents/shows/autism.world/notebooks/sue/notebook.html and https://www.amazon.co.uk/Reason-Jump-voice-silence-autism/dp/1444776770).* Sue and Naoki are both minimally verbal, so you will have to imagine that they are communicating by typing their thoughts on a keyboard.

N: Hey Sue, have you heard? Scientists claim they have developed a pill to cure autism. Here it is: we could actually try it and see if it works.

S: Wow, this is interesting…

N: I don't know… I used to think it'd be the best thing if I could just live my life like a normal person, but now I am in doubt whether I should take the pill.

S: Why did you change your mind?

N: Well, autism is such an inseparable part of our identity. Wouldn't eliminating it (if this is even possible) mean eliminating who we are?

S: Well, the possibility that I could be very autistic for the rest of my life always upsets me, so I would love to hear more about this pill.

N: Really?

S: Yeah, you see, people with 'high-functioning' autism are different, but basically they are just a variation of the norm. Some even have exceptional abilities that should be celebrated and encouraged. But people with severe autism, like me, are just trying to get through the day without hurting, tapping, flailing, biting, screaming, etc. It is a constant struggle. For instance, when I have to mask autism in class it takes a tremendous amount of effort. So I think that a cure for autism, would be wonderful.

N: But isn't it a struggle for everyone? Every human being, with or without disabilities, needs to strive to do their best; and for us having autism is normal, so we cannot know for sure what 'normal' is even like… I think I might well choose to stay as I am.

- <u>*Genetic essentialism:*</u> Currently, there is no cure for autism. Should we welcome genetic research to develop a drug to cure autism? Why? Prompts:
  - *If participants are in favour of a cure:*
    a) *Eliminating vs ameliorating:* A cure that eliminates autism altogether, or that helps autistic people get rid of the difficulties that they might experience because of autism?
    b) *Normalization:* For all cases of autism or only for severe cases of autism? Why? Who should decide which autistic traits should be cured? How can we decide which autistic traits are 'normal'? What is 'normal'? Who should decide which autistic traits are 'normal' and which traits should be cured?
    c) *Stigma:* What do you think may be the positives and negatives of eliminating all or some autistic traits for autistic individuals and for society in general? How could a cure for autism influence society's perceptions of autism and how society treats people with autism?

- *If participants are against a cure for autism:*
    a) *Compare and contrast:* Should we welcome genetic research to develop better treatment for cancer? Why? How is a cure for cancer different from a cure for autism?
    b) *If someone says that cancer is a life-threatening condition, while autism is not:* Should we welcome genetic research to develop a drug for a physical issue that is not life-threatening, such as hearing loss in deaf people? Why? Do you see a difference between using genetic information to cure disabilities that affect bodily traits and disabilities that affect traits associated with people's identity?

- *Genetic reductionism and resource prioritization: Participants will be given one set of magnets, with the following items written on: genetics (genetic causes of ASD); neuroscience (brain, cognitive capacity, hypersensitivity); treatments & interventions (behavioural, medical, educational, technology-based); services (projects on community inclusion, employment, family wellbeing, practitioner training); societal issues (ethics, sociological study, ethnography), adapted from (Pellicano et al., 2014, p. 757). They will then be told:* On these magnets you can read some areas of research that is currently conducted on autism. Imagine that you have a set amount of money that you can spend on these research areas. As a group, please rank these areas from the one you would spend the biggest amount of money on to the one you would spend the least amount of money. Please explain your choice.

  Prompts:

- If you think back of our discussion on the causes of autism and of the difficulties that autistic people encounter, do you believe that genetic research on autism should be prioritized? What could be the objection of the neurodiversity movement to such prioritization?
- Can you think of any other service in which families with autistic people may want to see funding spent on?
- Who should decide where money should be spent on?

Activity 2: Attitudes towards preventing autism

Genetic determinism and essentialism

The researcher will ask someone in the group to read a short online article in which Charlotte, a mother of two autistic children and one neurotypical child, presents her perspective on prenatal genetic testing for autism ('Prenatal tests and autism - a mother's story' by Charlotte Moore, The Guardian, 12 Jan 2009 https://www.theguardian.com/lifeandstyle/2009/jan/12/prenatal-autism-test).

Adapted from *Prenatal tests and autism – a mother's story*, by Charlotte Moore, *The Guardian*, 12 January 2009:

When my eldest son, George, was born, I had no way of knowing that he had an invisible lifelong condition that, 19 years later, would keep him almost as dependent as a toddler. George is autistic. So is his brother Mark, born 22 months later. Knowledge of autism has galloped forward since their early days. Soon a prenatal test may be available, to indicate whether a foetus is likely to develop autism. I'm glad that test wasn't available to me.

My first pregnancy ended in a termination. A mid-term scan showed the baby had no limbs. Such a child, I thought, would have no option but to be a hero. I considered this an unfair burden, and I ended his life, with great sorrow but without regret. It would seem logical, then, that I would have made the same decision about an autistic foetus. And yet there is no part of me that wishes away my sons' lives, or the life I have with them.

I have a third son, Jake, 10, who does not have autism. When I outlined the abortion debate, he was upset. 'George and Mark aren't sad about being autistic because they don't know they are,' he said. 'Anyway, what's wrong with being autistic?' Jake accepts the life he's always known. His brothers break his possessions, gobble up his chocolate, render the lavatory unfit for use, but he doesn't blame them. They're autistic; that's what they do.

Many articulate autistic people would argue that Jake's right; there's nothing intrinsically 'wrong' with being autistic, it's just that our non-autistic society does not meet their needs. Unlike my sons, who I believe have no perspective on their condition, the more able often suffer from awareness of their differences. I doubt that a prenatal test would be able to distinguish between severe autism and the 'high-functioning' kind, but in any case it's a mistake to think that life is easier for the more able. However, would able autists agree that their potential suffering should have been terminated before birth? I suspect not.

Most autists are physically healthy. A minority have epilepsy, and gut problems are quite common, but, for most, 'quality of life' depends on quality of care and understanding, appropriate education, and a living environment that takes account of their sensory hypersensitivities. Such conditions are achievable – but at considerable cost to the carers, usually the parents. Not only is the caring hard work, but there's no cut-off point. My friends' children are away on gap years, starting university, finding partners. George and Mark are giant children, and my responsibilities towards them will continue until I die.

A prenatal test without a termination would have prepared me a little for what lay ahead. But the thought that I might well have opted for termination makes me shudder. Abortion has always happened, and it always will. When the prenatal test is introduced, it will make no sense to decide

that a mother can choose to terminate, say, a Down's baby, but is obliged to keep a potentially autistic one. It's inevitable that many will be terminated.

Autism often means sleeplessness, eating problems, self-harming, aggression, destructiveness, bizarre behaviour of all kinds. It can destroy marriages, and it certainly doesn't help your finances. I would never condemn a mother who decided that she couldn't cope with these possibilities. But autism also means unique perceptions, special talents, weird humour, a view of the world untainted by greed, envy, malice, vanity, ambition. A society that aims to remove all the variables that make human life so fascinatingly complex is not a society I want to live in.

- *Prenatal testing and termination:* Do you agree or disagree with Charlotte's view? Why? What may be the arguments in favour and against Charlotte's view? Prompts:
- *Genetic essentialism:*
    a) *Impact of autism on families:* Charlotte writes: 'I would never condemn a mother who decided that she couldn't cope with these possibilities.' What does this tell you about one of the reasons why some parents may want to terminate their foetus that is at increased risk for autism?
    b) *Flourishing, good autistic life, non-identity problem:* How do you think the neurodiversity movement would argue against terminating the foetus? Do you think that autism is likely to decrease people's quality of life so much that it would be better for them not to be born?
    c) *Unique culture argument; loss of valuable diversity*: Charlotte says: 'A society that aims to remove all the variables that make human life so fascinatingly complex is not a society I want to live in.' Do you agree/disagree? What might the availability of these interventions at a very early stage say about the value that society attributes to autism and autistic people (e.g. in terms of tolerance, diversity, stigma etc.)? Do you think Charlotte is saying that she would not terminate or that there should not be an option to do a genetic test for autism on a foetus? Do you think these two arguments are different? Do you agree with any of them?
    d) *Compare and contrast*: How would your response change if the case was about deafness/paediatric cancer/heart disease rather than autism? Would you have the same/different concerns if the case was about deafness/paediatric cancer/heart disease rather than autism?

- *Genetic determinism:* Charlotte says: 'I doubt that a prenatal test would be able to distinguish between severe autism and the "high-functioning" kind.' *The researcher will remind participants that autistic symptoms are across a spectrum.* How does this information change your opinion/view?
- <u>*Preimplantation genetic diagnosis*</u>: *This example will be given particularly if participants are against prenatal testing because they are against abortion.* Imagine that a couple who already has two children with autism is planning to have another baby. Their geneticist tells them that they can access a procedure by which they will be able to select an embryo that has a lower risk of a genetic predisposition to autism, before implanting the embryo into mum's uterus. Should this couple be allowed to do so to prevent giving birth to an autistic child? Why? How is this different from Charlotte's case?
- *Compare and contrast:* How would your response change if the couple wanted to select in favour of the embryo that has a higher chance to develop autism? Should they be allowed to do so?
- *Sex selection:* One major risk factor for autism is being male. If parents simply decided to select a female embryo, would there be more/fewer/same ethical concerns as testing an embryo for autism risk and then selecting a lower-risk embryo?
- <u>*Obligation vs parental autonomy*</u> *(J. Harris, J. Savulescu): The researcher will show participants a piece of paper with the following quote.* A famous philosopher once said that, 'out of the possible children they could have, parents have a moral responsibility to select the child who is expected to have the best life, or at least as good a life as the other possible children' (Savulescu, 2001). Do you think that if a couple can identify that their embryo/foetus has a higher risk of autism, they have an obligation to eliminate that risk? Or do you think it is within parents' right to choose what to do? Why? Prompts: Do you have anything to say about 'the best life'?
    - *Compare and contrast:* How would your response change if the case was about deafness/paediatric cancer/heart disease rather than autism? And if the couple wanted to select in favour of the embryo that has a higher chance to develop autism?

Activity 3: Attitudes towards intervening early to reduce the impact of autism

Genetic determinism and essentialism

If you know that your child is at an increased risk to develop autism because they carry certain genes associated with ASD, then you could monitor their behaviour, you could keep an eye on

him/her. In case the child started to show symptoms of autism, you could enrol him/her in early intervention programmes such as 'speech therapy or social skills training or some sort of behavioural approach' (Simon Baron-Cohen, The Guardian 2014).

- *Testing for early intervention:* Should we genetically test babies for this purpose? Why? Prompts: Every baby or at-risk babies only?
- What may be the medical and psychological benefits and harms of knowing that a child is at risk of developing autism at a very early age, perhaps before they have showed any symptoms?
  - *Impact on the family:*
    a) *Genetic determinism and parental anxiety:* How could this information psychologically benefit/harm the child's parents?
    b) *Impact on family's understanding of the child:* How could this information impact on the way parents see and treat their child? How could this impact their child's development?
    c) *Disrupting family relationship:* How could this information impact the parents' relationship?
  - *Impact on the child:*
    a) *Early psychiatric label, stigma, sense of self:* How could this information benefit/harm the child's sense of self?
    b) *Early labelling vs right to an open future:* Even if the child's risk to develop autism was high, at present genetic testing would not provide any indication regarding the severity of the difficulties that the child might experience; there is no way to know whether they will experience a mild or severe form of autism. How does this affect your view on testing babies for their predisposition to autism?
- *Enrolling before symptoms:* There is currently much research conducted on infant siblings of people with autism. This is because we know that they are at increased risk to develop autism, since they share part of their genetic makeup with their older autistic siblings. In some of these studies, 'baby sibs' are genetically tested to see if they carry the same genes associated with autism as their older siblings; if so, they are enrolled in early interventions (e.g. interventions to improve their social communication skills), which are meant to reduce the risk that the baby sib will develop autism as their older sisters or brothers.

Should we enroll baby sibs in early intervention programmes before they have showed any symptoms of autism, on the basis that genetic test shows that they are at increased risk? Why?

a) *Impact on the baby sibling:* How could the enrolment benefit/harm the baby sib's sense of self?

b) *Impact on the sibling with autism:* What could be the psychological impact on the sibling who has autism?

c) *Genetic essentialism:* What might the availability of early interventions say about the value that society attributes to ASD and autistic people?

d) *Compare and contrast:* Couples' risk of having subsequent children with autism is 10-20% if they already have one autistic child and 30-50% if more than one. Would you be in favour or against enrolling the baby sib in an early intervention programme based on this information, if no genetic test is performed on the child?

Concluding questions

- Is there anything else that you would like to say?
- Is there anything that we have not covered?
- Thank you very much for your time.

Appendix I: Leaflet on neurodiversity

Autism and Neurodiversity

If you or someone in your family has received a diagnosis, or identifies as on, the autism spectrum, you might be interested in knowing more about the *neurodiversity movement*. The neurodiversity movement can provide a source of support and information about living with autism.



WHAT IS NEURODIVERSITY?

Neurodiversity is the differences in people's brains that affect how we experience the world.

The neurodiversity movement (NDM) argues that forms of 'neurodivergence' such as autism, dyslexia, and ADHD are inherent and valuable parts of the spectrum of human variation. There is no 'right' or 'wrong' way to develop. There is no 'normal' brain.

THE SOCIAL MODEL OF DISABILITY

Ideas about Neurodiversity parallel **the social model of disability**, which recognises that the negative consequences of having an impairment – i.e. the disabling quality of a condition – are often caused by societal structures rather than the condition itself.

For example, some autistic persons may react badly to lights or sounds that are common in the social world as they may overstimulate their senses.

The social model of disability argues that barriers in society create disability, and require reasonable adjustments for those who need them.

There are parallels between the neurodiversity movement and other social justice movements like the broader disability rights movement and feminism.



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Autism and Neurodiversity
Exploring Diagnosis

wellcome

The Neurodiversity Movement



WHO IS INVOLVED?

People like you:

- Autistic people and others discriminated against for their differences.
- Family members and other 'allies'.
- The movement works to eliminate oppression and stigmatisation and to obtain rights and support for those affected.
- Many of the most vocal activists in the NDM have come from the autism rights movement.

WHAT ARE THEY SAYING?

- Many in the movement see their conditions as an aspect of their identity that brings strengths as well as challenges, not as a disease to cure.
- Many in the movement welcome support and access to services that help with everyday life.
- They argue that children should not be subject to forced attempts to 'normalise' them.
- Consequently those in the NDM promote **acceptance of difference** and **valuing of strengths**.



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Comments? We welcome feedback:
blogs.exeter.ac.uk/exploringdiagnosis

RESOURCES

Blog posts

- *'Autism Resources'* by Kit Mead
- *'Welcome to parenting within the Neurodiversity Paradigm'* by Ally Grace
- *'Here's What Neurodiversity Is – And What It Means for Feminism'* by Cara Leibowitz
- *'Neurodiversity: Some Basic Terms & Definitions'* by Nick Walker

E-books

- *Being Autistic* by AutAngel
- *Welcome to the Autistic Community!* by the Autism NOW Center and the Autistic Self Advocacy Network

Online communities

- Parenting Children with Autism with Love and Respect
- The National Autistic Society

Online resource links

- **www.exeter.ac.uk/exdx/resources/feedback**
- **blogs.exeter.ac.uk/exploringdiagnosis**

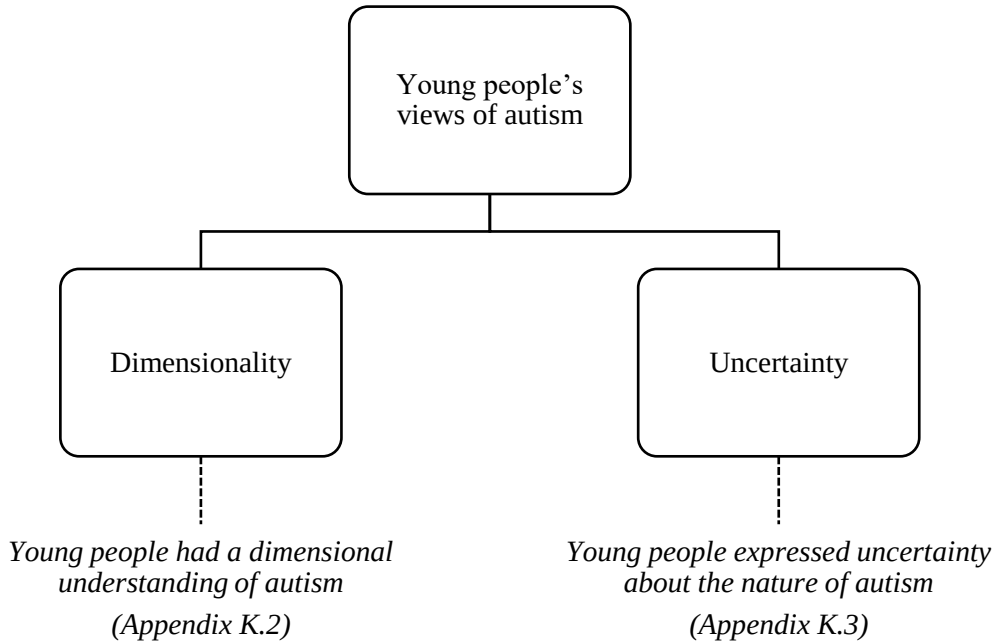
Artwork via the Art of Autism

Appendix J: Transcription conventions

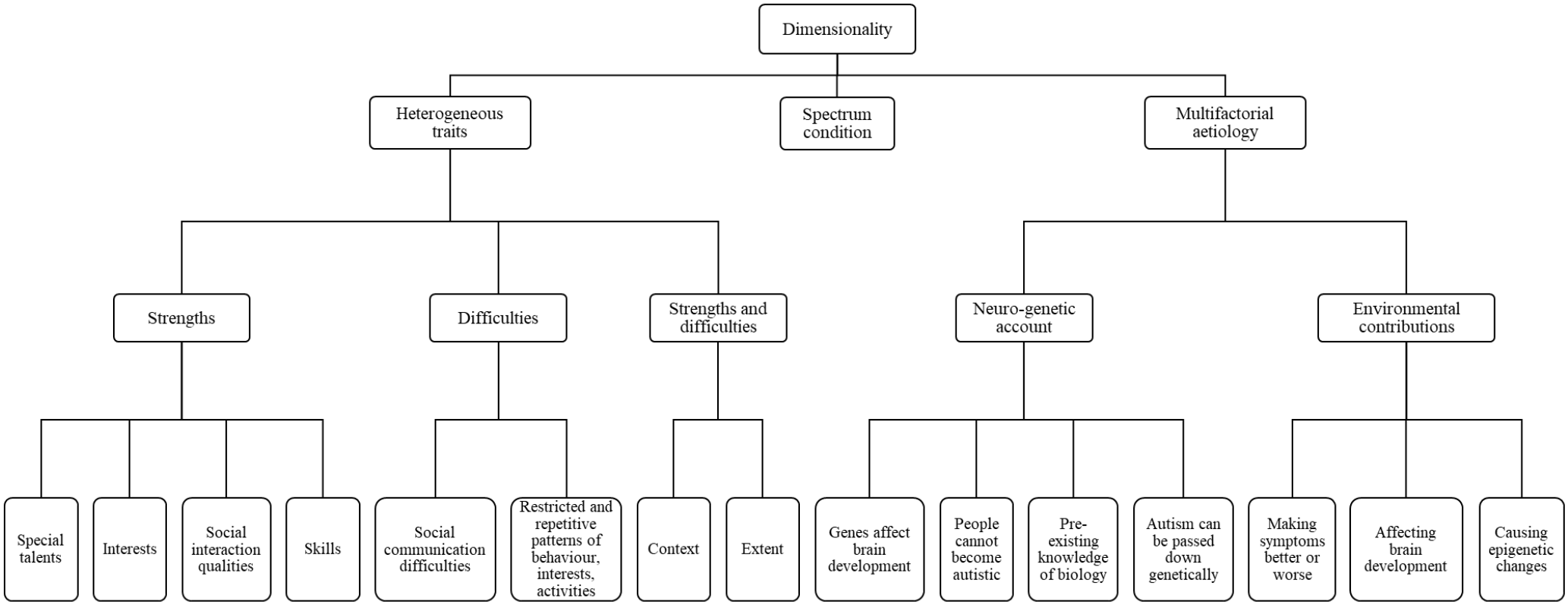
Symbol	**Explanation**
[ ]	Point at which a speaker is interrupted by another speaker
[word]	Suggestions regarding uncertain transcription
[unclear]	Unintelligible speech
word	Speaker's emphasis
…	Speaker pauses
[word]	Clarification of something said in a quote
[…]	An irrelevant part has been cut out
(FGDX)	At the bottom of a quote, it indicates the code of the FGD from which the quote is taken

Appendix K: Coding structure, FGD Part 1

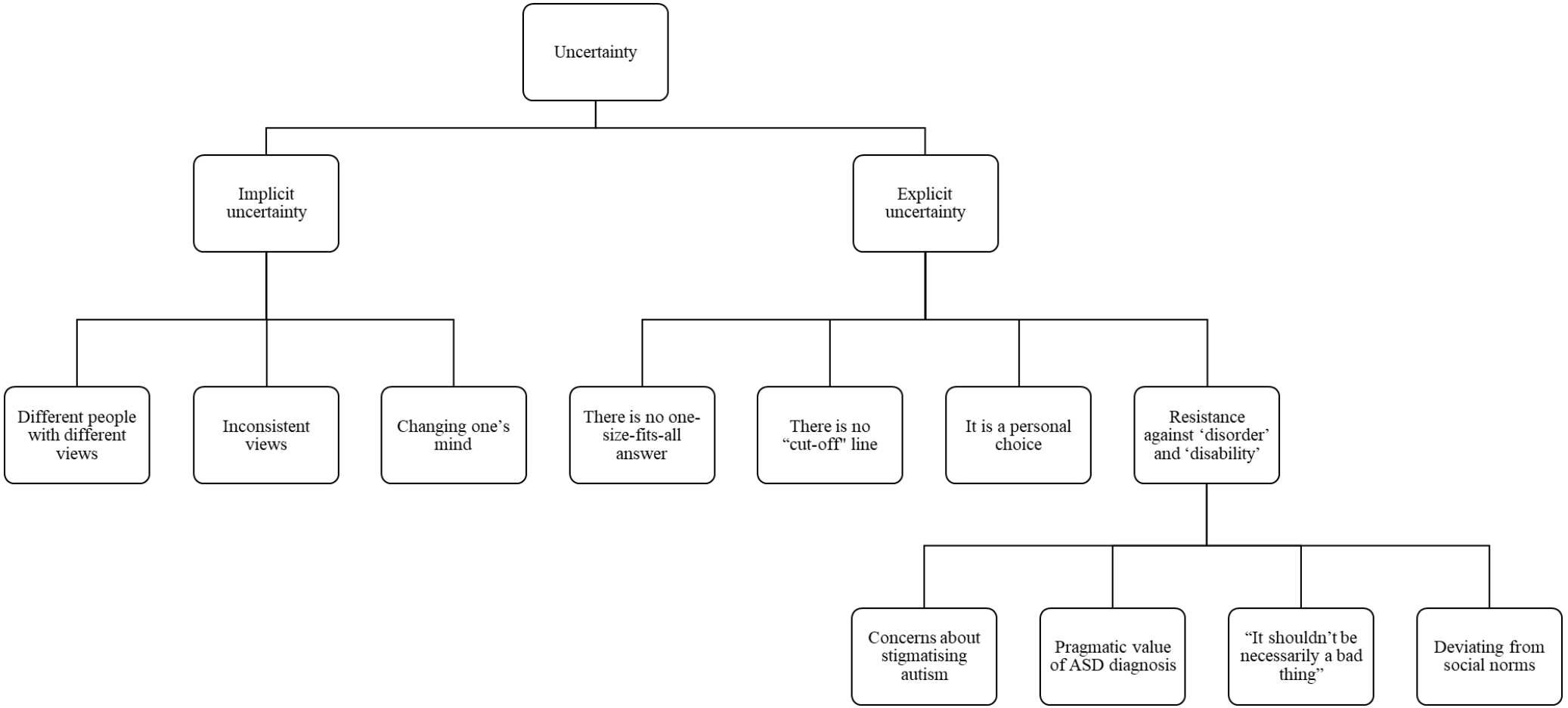
Appendix K.1: Overview of young people's views of autism



Appendix K.2: Young people's dimensional understanding of autism

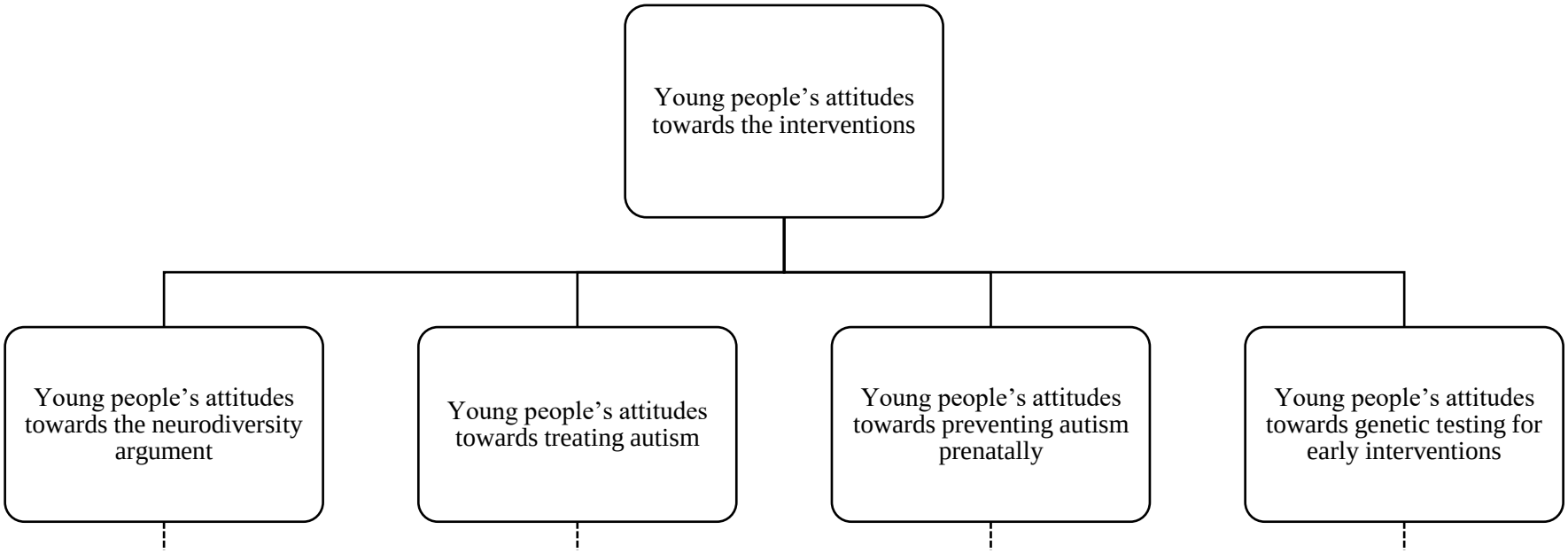


Appendix K.3: Young people's uncertainty about the nature of autism

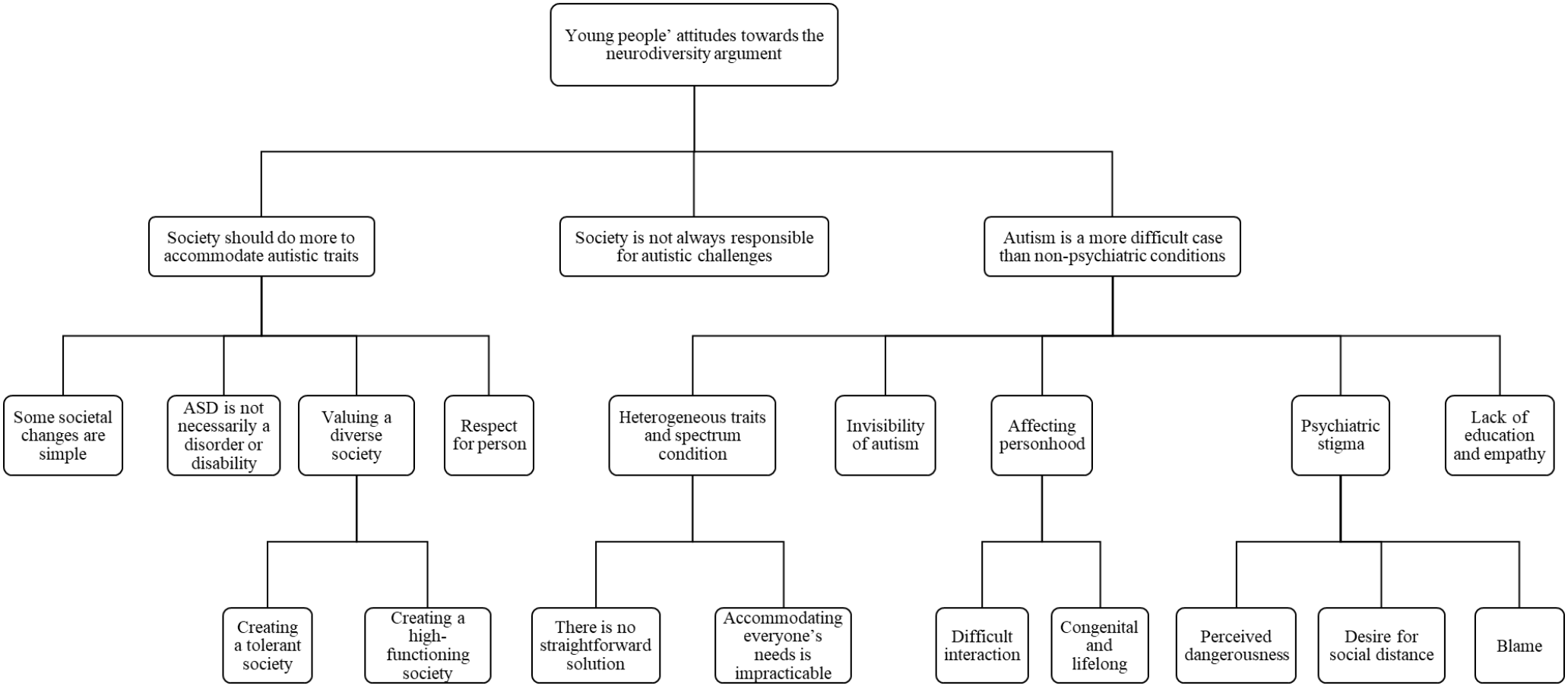


Appendix L: Coding structure, FGD Part 2

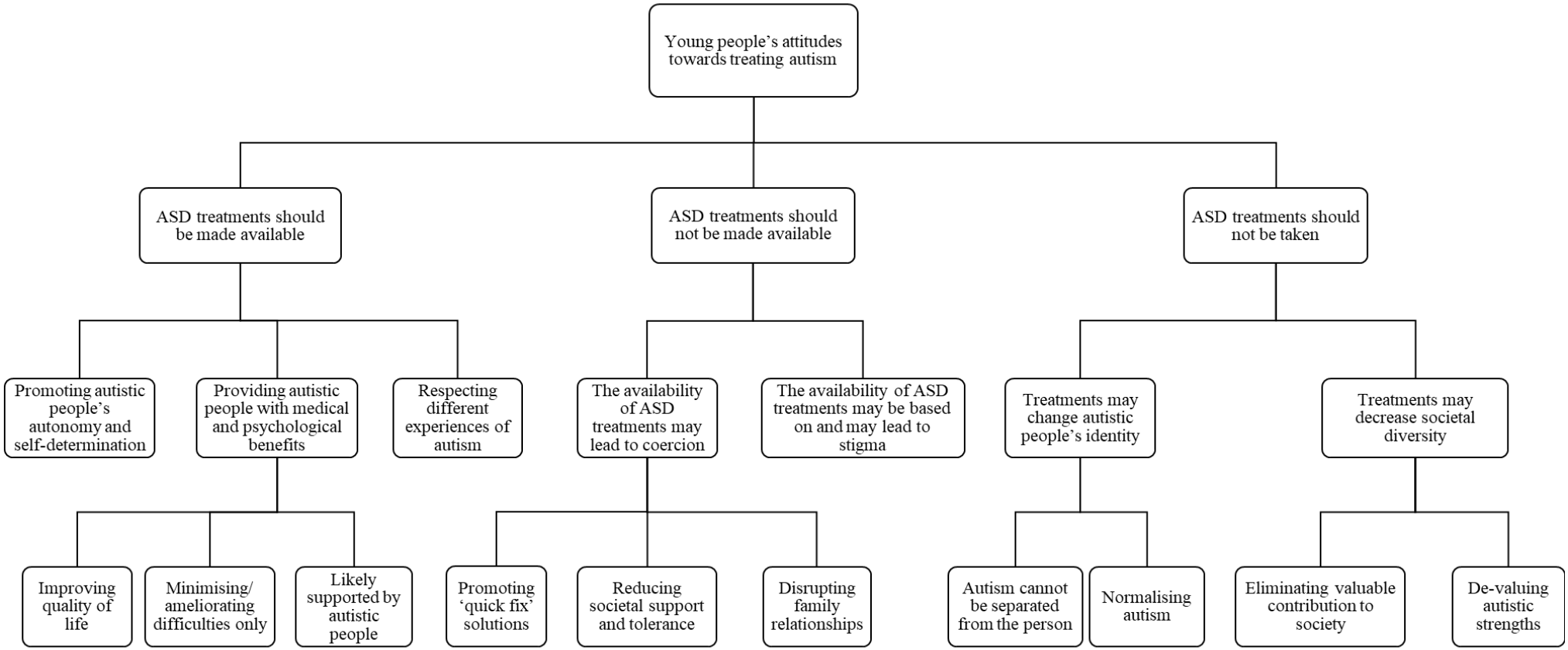
Appendix L.1: Overview of young people's attitudes towards the interventions



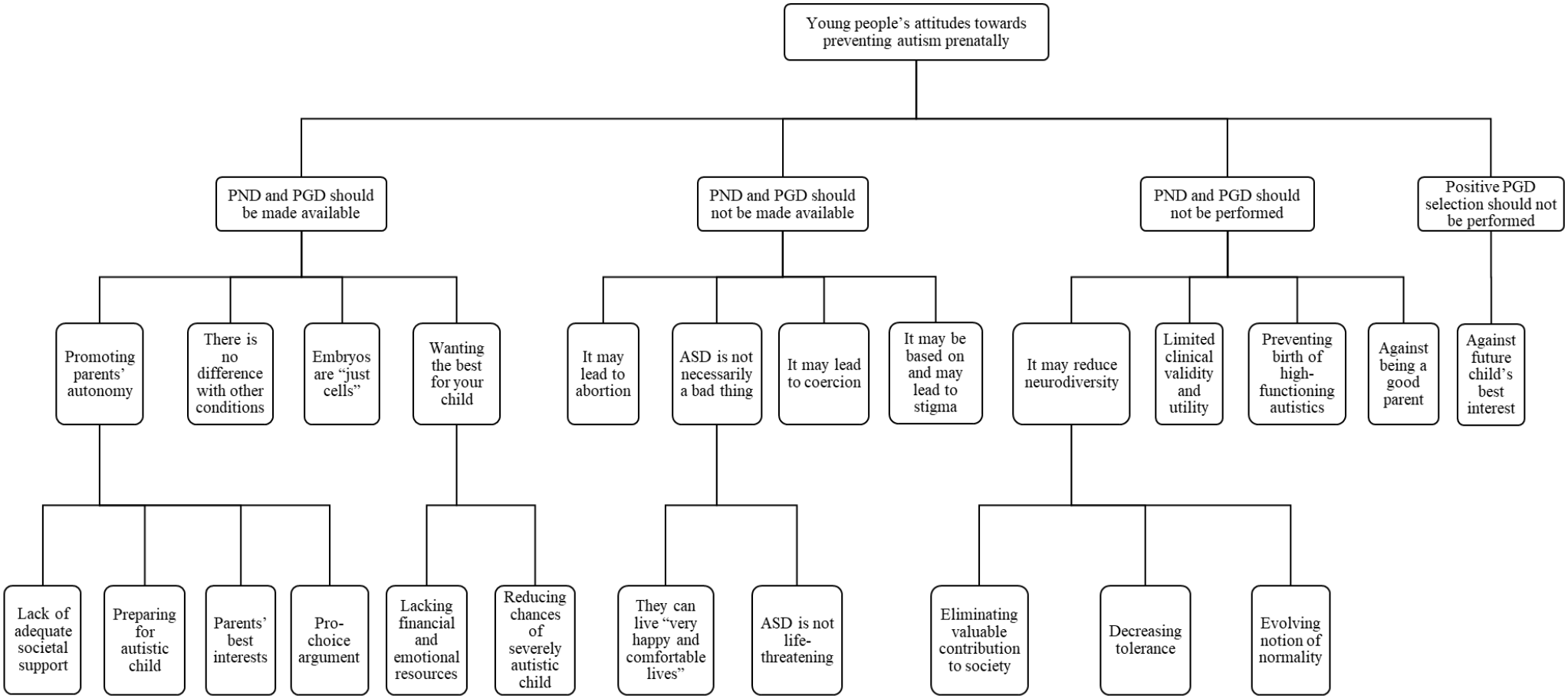
Appendix L.2: Young people's attitudes towards the neurodiversity argument



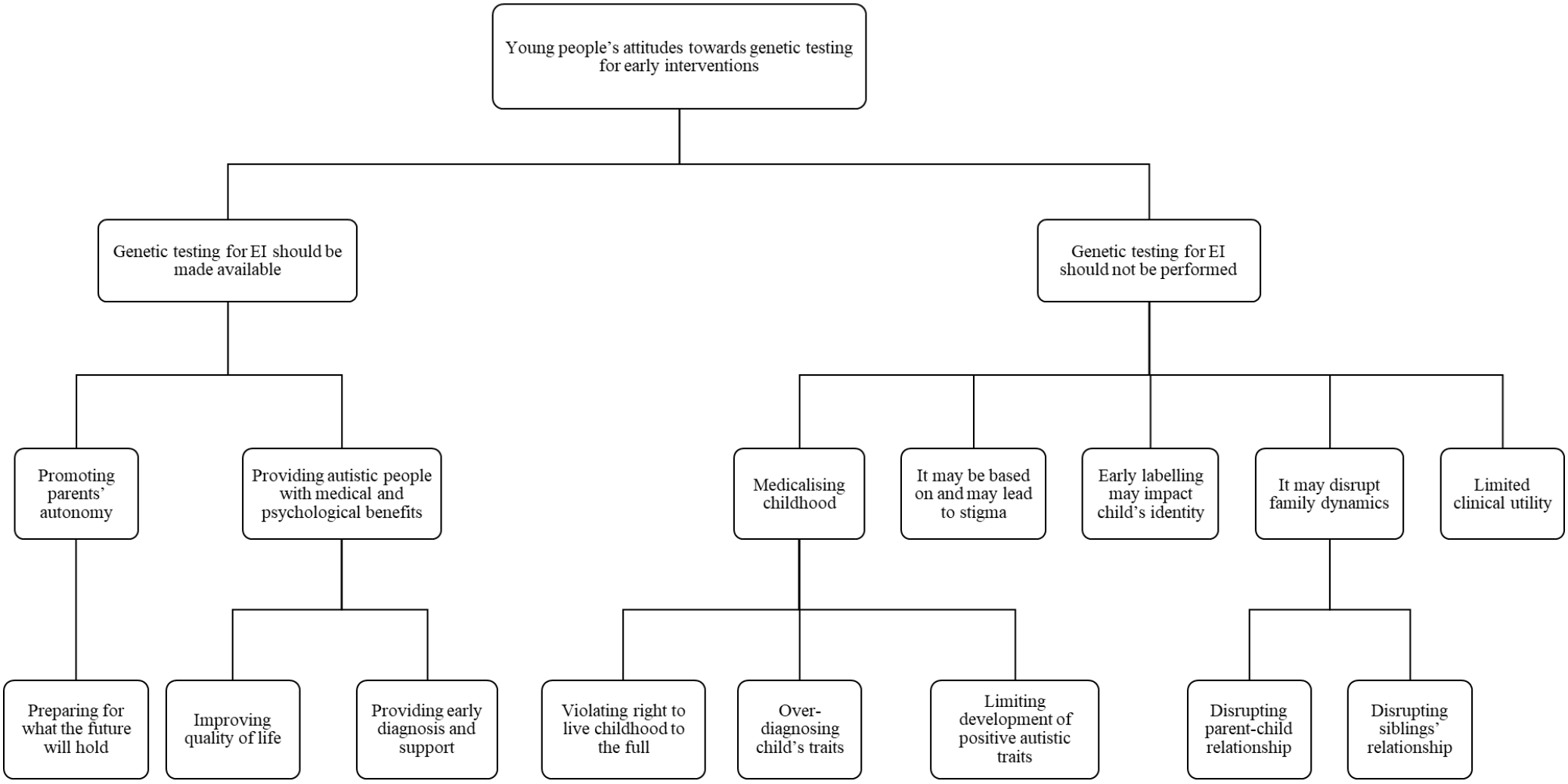
Appendix L.3: Young people's attitudes towards treating autism



Appendix L.4: Young people's attitudes towards preventing autism prenatally



Appendix L.5: Young people's attitudes towards genetic testing for early interventions



Appendix M: Sam's profile

Sam was diagnosed with autism when he was two. When he was a baby, he did not respond to his name or look at people in their eyes. He did not like to be hugged and often reacted in distress to new activities. Sam struggled with eating a varied diet, and he preferred the same food on the same plate every day. Sam had difficulties sleeping and sometimes would not sleep at all during the night. Sam is now 16 years old, and has severe learning difficulties so he attends a special school with kids who are similar to him. Sam cannot speak; however, he has some vocalizations and is being taught how to sign and use picture cards to communicate basic things like toilet or iPad. Sam enjoys the structure and routine at school, and particularly likes trampolining.

When Sam is not at school, he requires constant support and supervision from his parents for everyday activities, such as washing and getting dressed. This supervision is very important as Sam does not have an understanding of dangers around him. Sam's parents also cook a special diet for him, because he has always had gastrointestinal issues which make eating difficult for him.

When Sam gets distressed he cannot say it, and not being able to say it makes him more anxious. This means that sometimes he self-hits or bangs his head against the wall. He is also more prone to violent behaviour as a coping mechanism, so sometimes he hits his parents. Sam's parents know that he does not do so because he wants to hurt them, but because he is trying to express that he needs something. However, lately they have found coping with this behaviour difficult, because Sam is becoming taller and stronger. Moreover, they always try to avoid crowded and loud places, such as shopping centres, which make Sam agitated; however, Sam's behaviour is often unpredictable, which means that he could get distressed randomly, so making sure that he does not hurt himself is very difficult and stressful.

Sam loves watching water flowing from the tap, as he finds this very relaxing. He now lets people he trusts hug him, and is very loving, happy, and funny. He watches videos on repeat and often shares funny clips with others.

Appendix N: Outcomes arising from the DPhil

Peer reviewed publications

Kretzschmar, K., Tyroll, H., Pavarini, G., Manzini, A. and Singh, I. (2019) Can your phone be your therapist? Young people's ethical perspectives on the use of fully automated conversational agents (chatbots) in mental health support. *Biomedical Informatics Insights,* 11, 1-9.

Manzini, A. and Vears, D.F. (2018) Predictive psychiatric genetic testing in minors: an exploration of the non-medical benefits. *Journal of Bioethical Inquiry,* 15(1), 111-120.

Pavarini, G., Lorimer, J., Manzini, A., Goundrey-Smith, E. and Singh, I. (2019) Co-producing research with youth: the NeurOx young people's advisory group model. *Health Expectations,* 22(4), 743-751.

Other publications

Manzini, A., Mortimer, R.E. and Singh, I. (2017) Parental responsibility in the context of neuroscience and genetics, by Kristien Hens, Daniela Cutas, and Dorothee Horstkötter. Cham, Switzerland: Springer International Publishing; 2017. 246 pp. *Cambridge Quarterly of Healthcare Ethics,* 26(4), 681-685.

Manzini, A. and Vitiello, L. (2019) La medicina predittiva e il dibattito etico sui test genetici. In: Viafora, C., Furlan, E., and Tusino, S. (eds.) *Questioni di vita: introduzione alla bioetica.* Milan: FrancoAngeli, 367-388.

McKeown, A., Mortimer, R.E., Manzini, A. and Singh, I. (2019) Is coercion ever beneficent? Public health ethics in early intervention and prevention for mental health. In: Cratsley, K. and Radden, J. (eds.) *Mental health as public health: interdisciplinary perspectives on the ethics of prevention.* London: Academic Press, 45-68.

Selected conference presentations

Manzini, A. (2017) *Ethics and neuro-genetic technologies in psychiatry: why should young people's perspectives matter?* Postgraduate Bioethics Conference 2017 'Individuals and populations', 5 September 2017, St Catherine's College, Oxford (UK).

Manzini, A. (2018a) *"Better health for all" or devaluing individuals with "bad genes"? Exploring adolescents' moral attitudes towards advances in autism genomics.* 14th World Congress of Bioethics 'Health for all in an unequal world: obligations of global bioethics', 6 December 2018, Bangalore (India).

Manzini, A. (2018b) *How should we use genomic information about autism? A qualitative study on young people's attitudes*. European Association of Centres of Medical Ethics 35th Annual Conference 'Ethics in action', 7 September 2018, Vrije Universiteit, Amsterdam (Netherlands).

Manzini, A. (2018c) *Young people's moral attitudes towards advances in autism genomics: Preliminary findings of an empirical bioethics study.* Institute of Medical Ethics Summer (research) Conference, 19 June 2018, Oxford (UK).

Selected prizes and awards

Manzini, A. Wellcome Trust Secondment Fellowship in Humanities & Social Sciences 'Ethics,

youth mental health, and infectious disease outbreaks' [221455/Z/20/Z], October 2020-April 2021.

Goriely, A. Hallowell, N., Manzini A., and MIcochova H. Wellcome Trust Enriching Engagement Small Award 'Me and my genome: What am I really made of?', October 2020-October 2022.

Manzini, A. Institute of Medical Ethics Postgraduate Conference Grant for abstract submitted to 14th World Congress of Bioethics, Bangalore, India, 5-7 December 2018.

Pavarini, G., Manzini A., Lorimer J., and Singh I. OxTalent Award (Honourable Mention) for the video clip "Would you trust a chatbot therapist?" developed by the NeurOx YPAG, BBC Tomorrow's World, and the Wellcome Trust, 13 June 2018. OxTalent celebrates digital projects at the University of Oxford.

Pavarini, G., Manzini A., Lorimer J., Almeida A., and Singh I. Oxford Department of Psychiatry's Public Engagement Award for the work undertaken with the NeurOx YPAG, 9 January 2018.

Manzini, A. Kids Brain Health Network Travel Stipend for 'Outstanding abstract' submitted to 2017 International Neuroethics Society Annual Meeting, Washington D.C., 10 November 2017.

Manzini, A. Wellcome Trust 'Society and Ethics' Doctoral Studentship [203329/Z/16/Z], October 2016-October 2020.

Selected public engagement experiences

Co-organizer, *Make your own 'g'nome* event, Wellcome Centre for Ethics and Humanities, The Oxford Science and Ideas Festival, Oxford, 19 October 2019

Co-founder and co-leader, Neuroscience, Ethics and Society Oxford Young People's Advisory Group, University of Oxford, Oxford, February 2017-January 2019

Co-organizer, *Can your phone make you happier?* workshop, Bangalore, 4 December 2018.

Organizer, *Postgraduate bioethics workshop: critically engaging the public*, Biomedicine, Self, and Society Centre University of Edinburgh, 14 September 2018.

Guest speaker, *Gen-ethics: discussing ethical issues in genetic testing*, Philosophy Society, Magdalen College School, Oxford, 12 May 2017.