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From clinical genetics to genomic-based public health screening programmes: duty-based ethics as a guide for responsible implementation

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A renewed focus on disease prevention has placed genomics firmly in the spotlight. Policymakers and health services across Europe are considering ways to facilitate disease prevention and early disease detection through population-level initiatives such as newborn genomic screening and polygenic risk scores. This commentary explores, through the lens of duty-based ethics, the ethical considerations in the design of genomic screening programmes. As genomic medicine becomes embedded in public health strategies, a robust ethical framework is essential to ensure that its promises are realised equitably and responsibly.

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INTRODUCTION

Genomic medicine has evolved rapidly over recent years, driven by advances in sequencing technologies and expectations of more precise and personalised approaches to healthcare. Across Europe, national health systems are exploring ways to integrate genomic information into population-level public health initiatives, such as screening programmes, to identify those at increased disease risk and facilitate preventative action. For example, in the UK, recent health strategies have outlined ambitions to develop a genomics-enabled population health service [1]. Although prevention is a foundational principle of public health and primary care (or community) practice, its increased prominence within the broader healthcare setting represents a recent strategic reorientation, with genomics as a key enabler.

Some narratives propose genomics as the solution to complex health challenges, escalating costs and healthcare demand, with its benefits framed within a promissory discourse of clarity, innovation and transformation [2–4]. This discourse is underpinned by health, political and economic motivations. In the UK, this includes ambitions to position itself as a leader in life sciences, research and innovation.

Up to now, the main use of constitutional genomic testing has been in rare disease and inherited cancer predisposition diagnosis, through identifying disease-causing genetic variants in individuals presenting with features of inherited disease, and subsequent cascade testing of relatives for a known genetic variant. Looking ahead, genomic data is predicted to inform risks of both rare *and* common diseases (such as cardiovascular disease and common cancers). Emerging population health initiatives envisage genomic applications will start at birth through newborn whole genome sequencing, with applications across the life-course, using tools such as population-based polygenic risk scores and pharmacogenomics to tailor prevention strategies and personalise treatments.

Integrating genomics into population-level screening programmes represents a significant shift from clinical diagnostic practice. Such a transition requires consideration of the broader ethical implications in a way that goes beyond the autonomy-centred focus offered by Beauchamp and Childress' four principles approach [5]. Public health ethics draws on multiple traditions, including utilitarianism, virtue ethics and deontological (duty-based) approaches. Recent work on public health ethics emphasises the central role of duties and obligations in shaping ethical practice and the development of public health codes of ethics, highlighting their value in guiding institutional and policy responsibilities [6].

We therefore use a duty-based framework here to clarify the moral obligations of policymakers, healthcare providers and public health authorities when designing DNA-based screening programmes, enabling attention to the responsibilities that arise at the intersection of clinical genomics and public health practice [6].

ETHICAL DUTIES IN THE POPULATION-LEVEL APPLICATION OF GENOMICS

Duty to balance benefits and harms

At a population level, genomic-based screening promises improved disease prediction, enabling more precise risk stratification, targeted interventions, and earlier diagnosis. Its implementation requires robust evidence of benefit, assessed against established screening principles and supported by health-economic analysis [7]. It is also important to consider how

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genomic-based screening may affect engagement with existing evidence-based screening programmes and wider public health initiatives, for example, newborn screening programmes. The psychosocial effects of being at increased genomic risk, often for many years and potentially impacting multiple generations, require careful attention, recognising that individuals vary in perceptions of, and tolerance for, risk [8, 9].

Such evaluation is challenging, since the predictive value of many genomic variants remains uncertain, even if the analytical validity of the test is high. Some single variants found in the general population have a much weaker ability to predict rare diseases than when found in individuals with a personal or family history, reflecting the likely influence of additional disease-modifiers [10, 11]. This has been highlighted particularly by studies estimating disease penetrance in biobank participants [12]. More evidence surrounding risk estimates of genomic variants in unselected populations is needed, alongside new clinical paradigms that consider both the penetrance of the condition and the overall harms and benefits of intervention pathways. Without this, identifying such variants in programmes like newborn screening may create "at-risk" groups without a clear understanding of the associated clinical course, potentially leading to prolonged surveillance or interventions with uncertain benefit and increased healthcare costs.

Polygenic risk scores (PRS), which estimate disease risk based on the combined effect of thousands of genetic variants, have been proposed as a tool for population-based stratification of common disease risk. Despite considerable enthusiasm, PRS capture only a proportion of total disease risk [13, 14]. To date, they have shown limited ability to accurately identify individuals who will develop conditions such as common cancers or coronary artery disease, and they do not differentiate between those likely to develop aggressive versus indolent disease [7, 13]. Moreover, presenting genetic risk information alone may compromise individuals' understanding of their overall disease risk and erode their sense of agency in managing their health. PRS may be incorporated into integrated risk models to improve the precision of risk estimates. Specific evaluation would be required for each condition to determine the degree to which the PRS adds value to overall risk stratification, alongside integration with modifiable behavioural risk factors and support for behaviour change [15].

Duty to reduce inequalities

The incorporation of genomics into universal public health screening requires careful consideration since certain groups are already underserved by genomic medicine. Barriers to genetic testing, such as trust in healthcare services, genomic literacy, cultural and religious beliefs, language, perceived costs, and fear, are likely to be equally as relevant in population settings [16, 17]. The establishment of population genomic screening programmes will require deliberate efforts to address barriers to ensure that disparities are minimised and addressed.

Yet, genomic datasets, upon which much of the understanding of genetic variants is based, are overwhelmingly made up of individuals of European ancestry [18]. Interpreting the pathogenicity of genomic variants in non-European populations is made more difficult due to the lack of population-based evidence [19]. PRS, too, has a far higher degree of accuracy in European populations versus other ethnicities, meaning that their clinical use today would differentially benefit some populations over others and may widen existing disparities in disease prediction, diagnosis and treatment outcomes [20]. Addressing this gap will require large-scale genomic data generation in underrepresented groups, the release of granular summary statistics from large GWAS datasets to enable ancestry-matched analysis and systematic evaluation of PRS performance in diverse clinical settings to ensure equitable utility [20].

Duty of efficiency

In the UK, a legal duty exists for the health system to operate 'effectively, efficiently and economically' [21]. Whilst genomic screening might be seen as the answer to bypass lengthy waits for clinical genetics services, the effectiveness and cost-efficiency of disease prevention at a population level must be evidenced in advance of a system-level rollout.

Efficiency considerations should incorporate the necessary infrastructure developments and workforce to deliver a genomics population health service, alongside the financial and climate impact of long-term genomic data storage. They should also consider the impacts on current system capacity, including new obligations to monitor patients labelled at increased risk of disease and an increased number of patients identified with indolent conditions.

Although, in theory, people identified with lower genetic risk could be offered reduced disease-specific screening, in practice, the public views screening positively, making withdrawal difficult [7, 22]. As genomic prevention becomes a policy priority, the opportunity costs of diverting resources from other health needs, and the potential for genomic programmes to integrate with or disrupt existing services, must also be considered.

Health is influenced by many factors beyond the individual, including wider determinants such as family and social networks, diet, access to income, good quality housing, education and employment opportunities, transport and an individuals' surroundings [23]. There is a risk that an over-reliance on genomic data to address complex, multi-factorial health conditions will detract attention from the broader social, environmental and structural determinants of health. Moreover, because genomics alone cannot achieve the prevention of such diseases, this overemphasis risks further entrenching existing health inequities.

Duty of community engagement and involvement

Community engagement is a cornerstone of ethical public health practice. While population-level genomic screening will require broad public engagement, it also necessitates targeted, sustained engagement with specific communities. Meaningful community engagement strategies can contribute to improved health literacy, reduced health inequalities and more equitable health outcomes, while also building trust and accountability [24]. Considering the longstanding inequalities in genetics-related research and healthcare, efforts to extend the benefits of genomic medicine at the population level should begin with meaningful engagement with historically and currently underrepresented communities [25].

While genomic screening is often presented as a transformative innovation, its legitimacy ultimately depends on alignment with the priorities and concerns of the public and the diverse communities it aims to serve, priorities that may not always coincide. In a context shaped by economic austerity, pressures on public services, data-privacy anxieties and political uncertainty, genomic health may not be viewed as an immediate priority for many. Without genuine engagement that addresses these competing realities, genomic public health initiatives risk appearing imposed or disconnected from lived experience, undermining trust, legitimacy and uptake.

Although patient involvement is essential, population-level approaches to common diseases should not be unduly influenced by paradigms or experiences derived from rare disease contexts. As genomic medicine moves into public health application, inclusive decision-making drawing on public health, primary care, local government, communities and the wider public, as well as clinical genetics, will be crucial to ensure ethically grounded, equitable and publicly accountable implementation.

CONCLUSION

In this commentary, we highlight the new nuances that arise as genomics is integrated into public health screening programmes.

Using a duty-based ethical framework, we emphasise the principal responsibilities of decision-makers, ensuring that factors of importance to the public are fully considered rather than overshadowed by economic or political outcomes.

Balancing population benefit against uncertainty in risk prediction, preventing the widening of existing inequalities, making efficient use of constrained public resources, and embedding meaningful community engagement must be treated as interconnected ethical obligations rather than competing aims. Without sustained attention to these duties, population genomic screening risks reinforcing structural inequities, diverging from public priorities, and diverting focus from the wider determinants of health. However, when approached with rigour, transparency, and ongoing engagement, population genomics has the potential to contribute responsibly to prevention strategies that support and strengthen core public health values.

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ADDITIONAL INFORMATION

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