

# Findings from qual research



# Methodology and sample



**Patients:** SMA Type 2 OR 3, Older than or 18yrs of age, between 7 months – 17 yrs of age when first experienced symptoms







**Caregivers:** Child has SMA Type 1 OR 2, less than 18 yrs of age when first experienced symptoms



**Both Patients and Caregiver's child:** Unable to walk without cane/crutch or other forms of assistance for no more than 10 steps

## \*Note:

-1 caregiver in Chile has two children with SMA: 1 child is on a clinical trial for risdiplam and the other child has been treated with nusinersen  
- 1 caregiver in Brazil has a child who is part of a clinical trial on risdiplam

Methodology		45 minute web-assisted telephone interviews			
Country		Argentina	Brazil	Chile	Total
	Patients	2	2	3	7
	Caregivers	2	2	1	5
SMA types					
SMA Type 1		1	2	-	3
SMA Type 2		2	2	2	6
SMA Type 3		1	-	2	3
SMA treatment experience					
	Nusinersen	2	2*	1*	5
	*Risdiplam		1*	1*	2
	Treatment Naïve	2	2	3	7
Fieldwork dates		TDIs conducted between: November-December 2020			

# Physical, social and financial impact of disease means patients and caregivers are both heavily restricted in their movements and quality of life

## Impact of SMA



### Motor Function

**Restricted movement** – Declining motor function skills vastly reduce independence; need assistance with wheelchair, unable to undertake simple daily tasks and pain from lack of movement or scoliosis

**Swallowing and speaking** – food has to be liquidized, struggle with saliva accumulating and choking, unable to communicate problems sufficiently

*"It's bothersome, you need help with everything. The problem is not being unable to walk, the problem is not being able to move your arms."*

**Patient, Brazil, SMA Type 2**



### Breathing

**Inability to breathe independently** from ventilator (SMA Type 1) – patients are unable to leave hospital and are vulnerable to pulmonary infection, and need to avoid the cold, cold foods and drinks

**Difficulty breathing** for SMA Types 2 and 3 can lead to fatigue and affects sleeping



### Social / Professional

Have to **pre-plan when they can leave the house** (maj. patients & caregivers), **family dynamics impacted** where siblings become jealous, lose touch with friends and family or parents leave (caregivers)

**Lack confidence** to pursue romantic relationships (maj. patients)

**Difficulty attaining any work**, fatigue restricts ability (maj. patients), sacrifice/leave job opportunities (caregivers)

*"He has professional help, there is a nurse who looks after him all day and then my husband and I look after him when we come back from work and at weekends, so about 4 hours a day."*

**Caregiver, Argentina, SMA Type 1**



### Financial

Need **extensive insurance** to cover treatment, caregiver parents are unable to work in many cases as **become full time carers** and **adaptations to the house** (bathrooms, hoists to move patient around) are costly

# Unmet needs for patients and caregivers center around a lack of adequate disease knowledge and support within both healthcare system and society further impacting QoL

## Unmet needs for SMA burden

FREQUENCY OF MENTIONS



### Emotional care from healthcare professionals

- Doctors need to offer **more support for the emotional impact** of the disease
- Lack of consideration on the **impact the diagnosis has on patient's support network**
- HCPs can cause trauma through **lack of knowledge**



### Centralized service of care

- **No co-ordination or centralized management of specialists** (maj. ARG) - **no single physician** to explain the disease at point of diagnosis
- Severe **lack of access and resources** to provide support



### Physical support / accessibility

- **Accessibility** limited / places are unavailable due to lack of wheelchair access and disabled toilets- want available information on accessible footpaths / disabled playgrounds
- Easier and **more coordinated support** with therapy in one location - exhausting to maintain
- Increased support as caregivers age - physical toll from lifting patient is felt



### Societal awareness

- Society and HCP **do not have detailed understanding** of the disease
- **Lack of support and access** to any form of help or treatment
- **Funding** from insurance or state often **does not stretch** to all that is needed

*"As for the health system, well there are simply too many pain points... The resources are so limited and the management is poor with too much bureaucracy."*  
**Patient, Argentina, SMA Type 2**

# Patients and caregivers would like a treatment that is less painful and easier to administer with improved efficacy both in terms of duration and on motor skills

## Unmet needs for treatment

FREQUENCY OF MENTIONS



### Administration

#### Less painful, complicated route of administration

- Nusinersen especially worrisome for patients as wrong administration could mean paraplegia
- Ideally no more tracheotomy and gastric tubes



### Fast and durable efficacy

#### Better long-term efficacy than current treatment options

- Needs to **stop disease progression**
- Rapid disease progression in first 6 months: need **faster onset of action** and **durable efficacy** over time esp. on motor function



### Motor Skills (specifically)

#### Desire greater **improvement in movement**

- Especially concerned with movement in arms
- Important aspect for more independency
- Ideally, enable to be able to walk freely



### Dual efficacy

#### Treatment that targets **both, symptoms and genetic aspects of the disease**

- Suggest treatment that could be used in older patients to restore motor function, reactivate atrophied muscles and amend associated genetic elements of the disease as well (min. caregivers)

# The most important attributes for treatment choice are a combination of both motor function and breathing function in order to obtain a sufficient QoL

## Most important treatment attributes for patients and caregivers

*\*Note: attributes were prompted in a 100 point allocation exercise*



### Breathing Function ★★★

- Most important attribute for **caregivers**
- Determines life or death for more severe SMA types
- No longer needing mechanical ventilation is life changing and means that some patients and their caregivers would be eased the burden of staying in hospital

*"The most important for me is to improve the motor function, this is what SMA has affected me most and to increase the life expectancy, this is my main worry of living with SMA."*

**Patient, Argentina, SMA Type 3**



### Motor Function ★★★

- Most important attribute for **patients, SMA Type 2 and 3 vs Type 1**
- Some patients acknowledge it sounds illogical to be ahead of breathing and life expectancy but perceive a better, and possibly shorter life with movement and independence more valuable to them than a longer life without these amenities

*"The more dependant the patient is on a mechanical ventilator, the more severe it becomes. A patient using a mechanical ventilator may die of respiratory infections, he may die due to stoppers clogging the part where the ventilation comes out, he may die due to equipment failures, may have hypoxia if the ventilation is not adequate... So, in my personal experience, my child's breathing is the one that worries me the most."*

**Caregiver, Brazil, SMA Type 1**



### Overall Life Expectancy★★

- Scoring highly for both patients and caregivers
- However not main focus as motor and breathing function both provide quality of life which is deemed more important
  - Therefore extended life duration falls behind potential efficacy improvements of treatment

Key: ★★★ Most important   ★★ Important   ★ Least important

# In contrast, when weighted against each other side effects and administration fall lower in the list of priorities given the context of disease severity

## Less important treatment attributes for patients and caregivers

*\*Note: attributes were prompted in a 100 point allocation exercise*



### Safety ★

- Maj. not concerned as drugs have been well tested before coming to market – seen as less important in context of severity of disease
- Min. (caregivers, Argentina) see as important in terms of long term implications (gene therapy) or after seeing bad reaction to treatment in other patients



### Side Effects ★

- Maj. perceive all treatments as having side effects – if decided to take the drug then have already assumed risks of side effects (headache, nausea, dizziness, fever)
- Min. mentions of concern over renal impairment – a fear of haemodialysis (caregiver)
- Less important in Brazil vs administration



### Administration ★

- If treatment is effective and safe then administration is seen as not important given the severe context of the disease (maj.)
- Oral administration is highly desirable for easing anxiety and at-home administration (esp. vs. Spinraza), however it is surpassed by other factors such as efficacy
- Less important in Argentina and Chile vs Side effects