

Scottish Survey (Dec 2017 – Jan 2018) re: impact of 5q SMA and views on nusinersen

1. Survey Reach

Views were sought from:

Those who have not had access to nusinersen:

- Parents of children age < 18 years with SMA living in Scotland
- Adults with SMA and their partners / unpaid assistants living in Scotland
- Other relatives of children / adults living with SMA living in Scotland
- Families living in Scotland who have been bereaved by SMA

Those who have had access to nusinersen:

- Families in the UK whose child with SMA has been treated with nusinersen (including bereaved)

This was via a direct mailout inviting participation from:

- 50 households known to SMA Support UK

	Number
Households related to child living with SMA	17
Households related to Adult living with SMA	6
Households bereaved by SMA	23
Household – N/K	4
Total	50

Households were related to:

	Child	Adult
Type 1	2	
Type 1/2	1	
Type 2	7	2
Type 3	3	1
Total	13	6

Invitations were also sent out via SMA Support UK social media channels and e-news mailing list and Treat SMA networks.

2. Responses

- 29 returns from households related to:
 - 12 children living with SMA (of whom two are siblings)
 - 7 adults living with SMA
- 5 returns from households bereaved by SMA
- 3 returns from households in Scotland where child has received nusinersen treatment
- June 2016 Scottish population: 5,404,700
- Prevalence of the condition is 1 – 2 every 100,000
- Population who have SMA estimated to be between 54 – 108

We heard views from households connected with 22 living with 5q SMA who have not been treated with nusinersen. This is the views of 20 – 41% of the population in Scotland who have SMA (adults who have SMA and parents of children who have SMA).

3. Full results - Children and Adults not treated with nusinersen – total 19

1. SMA Type

SMA Type	Numbers	%
1 / 2	2	10.5
2	9	47.0
2 / 3	2	10.5
3	6	32.0
Total	19	100

N.B. Age < 36 months = 2

2. Adult with SMA / parent of child with SMA's assessment of the health impact experienced due to SMA (Total 19)

Health Impact	Numbers	%
Contractures	12	63
Pain	10	53
Osteopenia	3	16
Bone fractures	7	37
Scoliosis	12	63
Acute breathing difficulties	7	37
Chronic breathing difficulties	4	21
Prolonged meal times	5	26
Fatigue with feeding	3	16
Choking	6	36
Constipation	11	58
Excessive weight gain	4	21
Insufficient weight gain	6	32
Weight loss	4	21

3. Adult with SMA / parent of child with SMA's report of the interventions for their SMA (Total 19)

Intervention	Has / has had /expressed need for this		Numbers out of the 19 waiting for this
	Total numbers out of 19	% of the 19 respondents	
Standing frame	5		
Walking frame	1		
Manual wheelchair	12	63	1
Powered wheelchair	12	95	
Wizzybug	6		
Specialist car seat	1		2
Specialist seating	10	53	
Specialist buggy	2		1
Orthotics	10	53	1
Lycra suit	2	53	
Spine orthotics	8		
Spinal rods / fusion	5	26	
Chest physio	4		
Postural draining	1		
Oral suctioning	1		
Other physio	16	84	3
OT	14	77	1
Assisted cough	2		
Nebuliser	2		
Oral suctioning	1		
Short term Non-invasive ventilation	3		
Night time NIV	2		
Mechanical ventilation / tracheostomy	0		
Mobile arm supports	1		
Tube feeding (ng or nj)	2		
Gastrostomy	1		
Wheelchair accessible vehicle	12	63	1
Adaptations to toilet and bathroom facilities	16	84	1
Specialist bed	13	68	
Hoist (mobile or ceiling track)	11	58	
Other home adaptations (eg bedroom, ramps, lifts	11	58	1
Assistive technology	5		

4. Adult with SMA / parent of child with SMA's assessment of the help needed for daily living. (Total 19)

(Note: two children are age < 3 years and have SMA Type 1 / 2 – their needs will increase compared to other children their age. Also, this combines all ages and types of SMA)

Task	Needs full help / a lot more help than for someone of this age	%
Washing	14	77
Dressing	15	79
Toileting	15	79
Transferring	11	58
Eating / drinking	12	63

5. Adult with SMA / parent of child with SMA's report of who requires night care

- **14 of the 19 (77%) have night care**

6. Adult with SMA / parent of child with SMA's report of who provides any unpaid care that is needed. (Total 19)

Relationship	Number	%
Parent	16	84
Grandparent / other relative	11	58
Friends	4	21
Partner	1	5
Son / daughter	1	5
Other	1	5

7. Adult with SMA / parent of child with SMA's report of what, if any, other caring responsibilities the main unpaid carer has. (Total 19)

Other responsibility	Number	%
None	2	11
Other children	11	58
Ageing parents	5	26
Other	5	26

8. Adult with SMA / parent of child with SMA's report of which, if any, of the main unpaid carers have had to give up work due to their caring responsibilities. (Total 19)

Given up work?	Main carer	Main carer's partner	Other family	Total %
Yes completely	6	1	1	42
Yes – dropped to part time	4	3		37

9. Adult with SMA / parent of child with SMA's (total 19), report of whether the level of paid support and interventions provided is enough for the unpaid carer to:

'Task'	No / Not really	%
Keep physically well	8	42
Get enough sleep	9	47
Keep well emotionally	8	42
Keep socially connected	8	42
Manage financially	8	42
Work the hours they wish	10	53

10. Adult with SMA report (5 replies) of whether the level of paid support and interventions provided is enough for them to:

'Task'	No / Not really	%
Keep physically well	2	40
Get enough sleep	2	40
Keep well emotionally	2	40
Keep socially connected	3	60
Manage financially	3	60
Work the hours they wish	3	60

11. Adult with SMA / parent of child with SMA's report of whether they wish to access treatment – for themselves / their child (Total 19)

Access treatment?	Number	%
Yes	16	84
No	2	11
Not sure	1	5

12. Adult with SMA / parent of child with SMA's report of their expectations of treatment and what this would mean to them (Total 19)

Expectation	Number	%	
Slow down / stop progression	10	53	
Improve strength / recover skills	9	47	Almost all were expressed as a hope, but qualified by 'a slowing / stopping of progression would be enough of a gain to make it worthwhile'

13. Adult with SMA / parent of child with SMA's report if they had heard anything that concerned them about nusinersen in any way (Total 19).

Concern	Number	%
Site of application	2	11
Long term effects unknown	1	5
Increased chance of respiratory infection and potential liver problems*	1	

*added to reply - 'But it won't stop me'.

14. Adult with SMA / parent of child with SMA's report of whether they think NHS Scotland should fund nusinersen treatment for everyone with 5q SMA Types 1, 2 and 3.

Of those who made a comment on this, three replies from parents of children said all children with SMA should have the opportunity. All other replies were that all with 5q SMA should have the opportunity for access or they specified access should be by those with SMA Type 2 or 3 as well as those with Type 1.

4. Parents of children living with SMA – total 12 children, 11 families

1. Type of SMA

Type	Number
1 / 2	2
2	4
2 / 3	2
3	4

2. Ages

Age	Number
< 36 months	2
3 - 12	6
13 - 17	4

3. Parent of child with SMA's assessment of the health impact experienced due to SMA (Total 12)

Note that age / SMA Type – which will have an impact on whether these impacts are evident – is not taken in to account in this summary

Health impact	Numbers	%
Contractures	6	50
Pain	4	33
Osteopenia	1	8
Bone fractures	4	33
Scoliosis	7	58
Acute breathing difficulties	4	33
Chronic breathing difficulties	2	17
Prolonged meal times	2	17
Fatigue with feeding	1	8
Choking	3	25
Constipation	8	67
Excessive weight gain	2	17
Insufficient weight gain	5	42
Weight loss	2	17

3. Parent of child with SMA's report of the interventions for their SMA (Total 12)

Note that age / SMA Type – which will have an impact on whether these interventions are needed – is not taken in to account in this summary

Intervention	Has / has had	Waiting	Turned down	% who have / express need
Standing frame	4		1	42
Walking frame	1			8
Manual wheelchair	8	1		75
Powered wheelchair	6			50
Wizzybug	3			25
Specialist car seat	1	2		25
Specialist seating	7			58
Specialist buggy	2	1		25
Orthotics	7	1		67
Lycra suit	1			8
Spine orthotics	3			25
Spinal rods	4			42
Chest physio	2			17
Postural draining	1			8
Oral suctioning	1			8
Other physio	11	1	1	100
OT	9	1		83
Assisted cough	1			8
Nebuliser	1			8
Oral suctioning				
Short term Non-invasive ventilation	2			17
Night time NIV	1			8
Mechanical ventilation / tracheostomy				
Mobile arm supports	1			8
Tune feeding (ng or nj)	1			8
Gastrostomy				0
Wheelchair accessible vehicle	5	1		50
Adaptations to toilet and bathroom facilities	9	1		83
Specialist bed	7			58
Hoist (mobile or ceiling track)	5			42
Other home adaptations (e.g. bedroom, ramps, lifts)	6	1		58
Assistive technology	2			17

4. Parent of child with SMA's assessment of the help needed for daily living. (Total 12)

Note that two children who have SMA Type 1 / 2 are age < 3 years – their needs will increase compared to other children their age. Also, that this summary combines all ages and types of SMA

Task	The level of help level required compared to the parent's expectation for a child of this child's age			Full help	Full help / a lot more help No. (%)
	As expected	A bit more	A lot more		
Washing	3		3	5	8 (67%)
Dressing	2	1	2	6	8 (67%)
Toileting	3		2	6	8 (67%)
Transferring	2		2	7	9 (67%)
Eating / drinking	4	1	5		5 (42%)

5. Parent of child with SMA's report of who requires night care (Total 12)

- 7 said 'yes' (58%)
- For 6 of the 7 who said 'yes' (50%), this was 4 or more times a night

6. Parent of child with SMA's report of the number of Health and Social Care Professionals they saw in 2017 seen in connection with their child's SMA (Total 12 replies)

- Range 5 – 17 professionals
- Average 10 professionals

7. Parent of child with SMA's report of their estimate of how many appointments they have each a month and much time they spend on these, travel time, chasing up equipment etc in relationship to their child's SMA (Total 12 replies)

- Range 2 – 6 appointments / month
- Range 2 – 80 hours taken up on SMA related tasks (not caring tasks)
- Six (50%) estimated over 20 hours / month with one as much as 80 hours
- Note - higher amount of time for those with younger child with SMA Type 1 / 2

8. Parent of child with SMA's report of their experience of being able to access the nursery / school / leisure options of their choice

	Never				Always
Nursery / school		8.33%	16.67%		75%
Social and leisure options		25%	33.33%	25%	16.67%

9. Parent of child with SMA's report of how many hours of paid care / week they receive to assist with their child. (Total 12)

- None for five of the children, including two who are under age 3 and one who is age 5-6. Two of these with no paid care have SMA Type 1 / 2
- One child has 6 hrs/wk; one child has 40 hrs/wk

10. Parent of child with SMA's report of who provides the unpaid care (Total 12)

Unpaid Carer	Number	%
Mother	10	83
Father	10	83
Grandparent	5	42
Other relatives	3	25
Friends	2	17
Other	2	17

11. Parent of child with SMA's report of other caring responsibilities of the main carer (Total 12)

Responsibilities	Number	%
None	3	25
Other children	9	75
Ageing parents	3	25
Other	2	17

12. Parent of child with SMA's report of who of the unpaid carers has had to give up work due to their caring responsibilities (Total 12)

	Main carer	Main carer's partner	Other family	Total %
Yes completely	4	1	1	50
Yes – dropped to part time	2	2		33

13. Parent of child with SMA's report (Total 12) of whether the level of paid support and interventions enough for the unpaid carer to:

'Task'	Yes	Not really	No	Don't know	Not really / no %
Keep physically well	4	3	3	1	33
Get enough sleep	2	3	4	2	58
Keep well emotionally	3	3	3	3	50
Keep socially connected	3	2	3	4	42
Manage financially	4	1	5	1	50
Work the hours they wish	3	2	5	1	58

14. Parent of child with SMA's report of their knowledge about nusinersen (Total 12)

- 75% report that they know a lot;
- 25% report that they don't know a lot but are interested

15. Parent of child with SMA's report of their wish for their child to access nusinersen treatment (Total 12)

- 100% Yes

16. Parent of child with SMA's report of their expectations of treatment and what this would mean to them (Total 12)

- 50% that it will slow the progress of the condition
- 50% that strength will improve and physical ability improve

17. Parent of child with SMA's report if they had heard anything that concerned them about nusinersen in any way. Their concerns about the treatment (Total 12)

One raised concerns about the site of the application and that long-term effects are unknown. No others expressed concerns

18. Representative comments (gender removed from text as far as possible to protect anonymity) from the replies

- **Parent of young person age 16 with SMA Type 2**

Our child's scoliosis has had a huge impact on their life both physically and emotionally. They ...suffer from anxiety mainly because of this.

Our child is fully reliant on help in all aspects of personal care. They are unable to wash dress shower or toilet themselves. This is extremely difficult for someone their age as they watch their peers getting on independently and this can be very emotionally distressing for them.

The biggest challenge in having a child with SMA is learning to adapt your life to meet the needs of your child not just the physical and emotional demands but the financial demands as well as anything needed for a child with a disability comes with a huge price tag.

Their bedroom feels like a hospital room to them with their bed hoist etc. They tend not to invite friends over due to all the equipment.

The main things is trying to help your child become a well-adjusted, independent, strong individual who accepts that they have to live with this horrible condition for life. SMA rules our life's for so many reasons and to watch your child suffer and deteriorate in front of your eyes is the most heart-breaking thing for any parent.

To have our teenager reach up to scratch their own head or to lift their hand to wave goodbye or to even move a little in their bed to get comfortable would mean more than life itself.

I feel all children with SMA should at least be given the opportunity to see if this drug can help in any way no matter how small because something small like brushing your teeth to a healthy person would mean the world to someone with SMA

- **Parent of child age 10 with SMA Type 2**

Despite spinal surgery at an early age (8) and a week in ICU post op the procedure was a huge success. Our child sits up straight in their chair and compared to other SMA2 peers without surgery the difference is marked.

1. Strain on parents' relationship
2. Lack of sleep. Six nights per month of respite isn't enough
3. Lack of free time for parents
4. Physical element is very demanding for both child and carers
5. Emotional aspect when you can see your child struggling with day to day tasks and getting progressively weaker
- 6 Child's inability to participate in so many activities and play with brothers/sisters and peers unless carer sitting next to them
7. Constant possibility of repeated chest infections and consequent reduced lifespan

Looking at the economics whilst treatment is currently expensive, the cost will decrease on pick up and in any case will be offset by subsequent reduced care needs and hospital stays.

- **Parent of child age 2 with SMA Type 2**

Physiotherapy is extremely limited. Sessions are once every two months (when is well know SMA children need it daily) and they only assess our child's deterioration or needs, but not really do much about it. There is no programme, no set of exercises or routines provided to the parents. We have to investigate on our own hoping that the sources of information are reliable. The care received is extremely limited, specially when it comes to physiotherapy.

Turning 4 times a night and the ventilation monitored up to ten times a night. My wife and I live by ourselves in the UK. We have no relatives here, so we always have to manage by ourselves.

We can hardly spend any time together as husband and wife, even with the simplest thing as watching TV together. We always have to take care of the needs of our baby by ourselves and spend countless hours trying to give them the care (physiotherapy) they should be receiving from professionals by ourselves.

We cannot decide to have more children, as our little one has so many caring needs. My wife cannot work because she has to dedicate her life entirely to our child. This has been emotionally draining for her as, instead of enjoying caring for our child, it has turned to be in a forced responsibility. I think everybody deserves the chance for a break.

Every day is a challenge. Waking up every night to help our baby rotate and sleep a bit better it's draining. We cannot even imagine their frustration, but I can tell you being sleep deprived every single day has been particularly horrible.

Please believe me, having a treatment that could help my child doing the simplest thing, even hugging me or their mother with at least the same strength than is in my own little finger, would be a huge blessing of them and our lives, Oh, I can't even begin to describe how challenging and messed up SMA. This is a condition that tears to the very soul.

Receiving nusinersen would mean the NHS could reduce the level of care given through life to these children.

- **Parent of young person age 17 with SMA Type 2**

The Lycra suit was a nightmare to get on as so tight. Spinal brace was very hot and uncomfortable to wear.

- **Parent of a young person age 11 with SMA Type 3**

Tiredness, backache, lack of time for myself , lack of time for other child, stress. My children lost their childhood. Lack of time for my other child, lack of places we could go together.

Better prevent. So many specialists involved, SMA requires loads of their time. Could save not only their time but also improve the lives of many people physically, mentally and emotionally.

- **Parent of a child age 8 with SMA Type 3**

The biggest challenge we face with our child is how to support them mentally and emotionally as they grow up.... Dealing with the condition physically will be hard but having to deal with the mental side has been harder

- **Parent of a child age 19 – 35 months with SMA Type 1 / 2 (specific age not given)**

Breathing issues makes our child very prone to chest infections. We avoid sick people and extreme weather conditions or situations which make them vulnerable. (Restricting them socially with friends)

At such a young age the biggest concern for us is our mental preparation for physical deterioration and the problems we will face as a family

These kids deserve to thrive, not to be left wasting away muscles where there is a known treatment to combat the deterioration of this debilitating condition.

Nusinersen gives us hope. The prospect of accessing this treatment creates a positive outlook on all our futures rather than being faced with a horrendous bleak future with this condition.

- **Parent of a child age 5 with SMA Type 2 / 3**

As they get older and bigger it becomes harder

As a parent it's hard to watch our child struggle and to see them decline in what they could do to now ... I see them weakening and become less capable but still achieving as much as they can ... I wish I could take away this SMA so they didn't have to struggle so much

SMA is a condition that robs children of their ability not just mobility they have to slowly feel themselves decline and become less able – it's hard to put into words, how to explain the upset when they see peers getting better and more independent they are getting weaker and more dependent along with the physical the mental and emotional upset - to have a sound mind but your body is failing is hard to deal with but as a child with sma there is nothing they can do.

I know it's not a cure but anything that could slow down or reduce the progression would be of benefit. As a parent mother it's hard to watch your child struggle and to see them decline in what they could do to now

- **Parent of a child age 9 with SMA Type 3**

Our child experiences extreme frustration and upset at the necessity for use of equipment and adaptations.

For our child:

Lack of independence for a child their age

Lack of ability to toilet independently

Lack of mobility and 'getting about'

Peer relationships and inclusion

Frustration and distress at the perceived unfairness of their condition

Limitations on their fulfilment of hopes, dreams, and potential

For the family

Taking time out of work to attend appointments

Constantly 'pushing' to get what our child needs / not feeling that we are doing enough

Emotional difficulties/distress and extra stress

Extra vigilance, worry and uncertainty about everyday activities and about what the future holds for our child

Challenging to help child be as independent as possible, and to fulfil their potential

Sibling relationship management

If a treatment could help in any way to delay or slow the rate of decline or maintain the motor function our child currently has then we believe this will have a significant impact on their quality of life, independence and mental health.

We believe that even relatively small physical effects of a treatment could have significant and impactful practical and emotional benefits for our child and family.

5. Adults with 5q SMA – Total 7

1. SMA Type

Type	Number	%
2	5	71
3	2	29

2. Ages

Age	Number
18-25	2
26-30	2
41 - 45	1
51 - 55	1
56 - 60	1

3. Adult with SMA's assessment of the health impact experienced due to SMA Health impact (Total 7)

Health impact	Numbers	%
Contractures	6	86
Pain	6	86
Osteopenia	2	29
Bone fractures	3	43
Scoliosis	5	71
Acute breathing difficulties	3	43
Chronic breathing difficulties	2	29
Prolonged meal times	3	43
Fatigue with feeding	2	29
Choking	3	43
Constipation	3	43
Excessive weight gain	2	29
Insufficient weight gain	1	14
Weight loss	2	29

15. Adult with SMA's report of the interventions for their SMA (Total 7)

Interventions	Has / has had	Waiting	Turned down	% who have / have express need
Standing frame	1			14
Walking frame				
Manual wheelchair	3			43
Powered wheelchair	6			86
Specialist seating	3			43
Orthotics	2			29
Lycra suit	1			14
Spine orthotics	5			71
Spinal surgery completed	1			14
Chest physio	2			29
Postural draining	1			14
Oral suctioning	1			14
Other physio	2	2		57
OT	4			57
Assisted cough	1			14
Nebuliser	1			14
Oral suctioning				
Short term Non-invasive ventilation	1			14
Night time NIV	1			14
Mechanical ventilation / tracheostomy				
Mobile arm supports				
Tune feeding (ng or nj)	1			14
Gastrostomy	1			14
Wheelchair accessible vehicle	6			86
Adaptations to toilet and bathroom facilities	6			86
Specialist bed	6			86
Hoist (mobile or ceiling track)	6			86
Other home adaptations (eg bedroom, ramps, lifts	4			57
Assistive technology	3			43

4. Adult with SMA's report of the help they need with daily living tasks (Total 7)

	Independent	Some help	Full help	Total needing Full / lot of help %
Washing	1	2	4	86
Dressing		2	5	100
Toileting		2	5	100
Transferring		2	5	100
Eating / drinking	2		2	29
Preparing meals		2	5	100

5. Adult with SMA's report of how many need night care (Total 7)

- **7 (100%)**

6. Adult with SMA's report of their estimate of the number of Health and Social Care Professionals they saw in 2017 in connection with their SMA (Total 7)

- Range 4 – 8 professionals
- Average 7 professionals

7. Adult with SMA's report of their estimate of how many appointments they have a month and much time they spend on these, travel time, chasing equipment etc related to their SMA each month (Total 7)

- Range 0 – 1 / 2 appointments each month
- Range 1 – 50 hours taken up

8. Adult with SMA's report of their ability to access opportunities (Total 5 replies)

	Never				Always
Education / work	20%		20%		60%
Social and leisure	20%	20%	20%	20%	20%

9. Adult with SMA's report of how many hours of paid care / week they receive

- Range: 0 hours for three adults – 91 hours/week.

10. Adult with SMA's report of how many hours paid health / social services funded care / week they receive

- Three people none
- One person 57 hrs plus 2 sleepovers
- Three people 70 – 91 hrs

11. Adult with SMA's report of who provides any unpaid care (Total 7)

Carer	Number	%
Parent	6	86
Other relative	3	43
Friend	2	29
Partner	1	14
Son/daughter	1	14

12. Adult with SMA's report of the other caring responsibilities of the main unpaid carer (Total 7)

Other responsibilities	Number	%
None	2	29
Other children	2	29
Ageing parents	2	29
Other	3	43

13. Adult with SMA's report of whether the unpaid carers had had to give up work due to their caring responsibilities (Total 7)

	Parent	Partner	Total %
Yes completely	2		29
Yes – dropped to part time	2	1	43

14. Adult with SMA's report (5 replies) of whether the level of paid support and interventions is enough for the them to:

'Task'	Yes	Not really	No	Not really / no %
Keep physically well	3		2	40
Get enough sleep	3		2	40
Keep well emotionally	3		2	40
Keep socially connected	2		3	60
Manage financially	2	1	2	60
Work the hours they wish	2	1	2	60

15. Adult with SMA's report of whether the level of paid support and interventions is enough for the main unpaid carer to:

'Task'	Yes	Not really	No	Not really / no %
Keep physically well	5		2	29
Get enough sleep	3	3	1	57
Keep well emotionally	5		2	29
Keep socially connected	5	1	1	29
Manage financially	2	2	3	71
Work the hours they wish	3		2	29

16. Adult with SMA's report of their knowledge about nusinersen (Total 7)

- 5 (71%) report they know a lot
- 2 (29%) report they don't know a lot but are interested

17. Adult with SMA's report of their wish for access to the treatment (Total 7)

- 4 (57%) Yes
- 2 (29%) No
- 1 (14 %) Not sure

18. Adult with SMA's report of their expectations of treatment and what this would mean to them (Total 7)

- 29% Stop progression of the condition
- 29% Increase strength
- 40% Any improvement to assist with independence

19. Adult with SMA's report of any concerns about anything they have heard about nusinersen (Total 7)

One expressed concern re: the site of application

One expressed concern about the chance of respiratory infection and potential liver problems but added, 'But this won't stop me'

No other concerns were mentioned.

19. Representative Comments (gender removed from text as far as possible to protect anonymity)

Adults with SMA Type 2:

- **Age 18 – 25 (specific age not given)**

I need care every 1 to 2 hours. I ask my Mom to come in to change a position of my legs and arms.

My scoliosis is the most irritating as I cannot access my wheelchair, I cannot go outside unless my Mom carries me into the car.

I have been watching many people around the world who are receiving this treatment and I can only see positive effects. I think everyone should have a chance to access this treatment regardless of their age or type of the illness.

- **Age 24**

My contractures could easily be eliminated by regular physio - which I don't always have access to because of lack of [appropriate support].

Growing up with spinal scoliosis was one of the most awful parts of my life, if I could undo it I would. Developing scoliosis meant pain, inability to breathe easily, eat properly and go to school without being in pain. Also, aesthetically scoliosis is not what is portrayed as the ideal shape of the spine - we can go back and forth re arguments but ultimately, I'd choose not to have that deformity.

Needing assistance is THE WORST part of my type of disability. I am an independent 24-year-old - I want to live my life exactly as I want and the biggest thing stopping me is my physical dependency on others. It impacts my life in a way you'll never understand if you don't have my condition.

- **Age 51**

Ensuring correct/enough funding is in place to support me living independently

Finding suitable PAs - recruiting can be a nightmare

Accessible holidays that don't cost a fortune

Finding suitable powered wheelchairs

I feel I have been more lucky in having adaptations done in my house, but know many who have to fight for them which is a basic human right

- **Age 44**

Having to wear back brace means needing the help of two people to put it on. It's physically challenging for those helping and also very impractical in terms of finding carers. Getting a suitable brace made is also very difficult. The Orthotics department seems to be the poor relation of the health service.

Needing full help with transfers and toileting is the biggest issue for me in all aspects - physically, emotionally and practically. I feel it's the one thing that if I didn't need help with it my life would be fuller and more independent.

If there was a drug/treatment that would just improve things enough, e.g. to be able to transfer myself to be able to go to the toilet more independently it would not only give me more independence and dignity but also cut down on the huge expense of carers who quite often, particularly at work, are needed for little else.

6. Parents of adults with SMA - Total 3

Three parents of adults responded to the survey.

Their replies and comments echoed those of their son / daughter.

- **Parent of person age 44 with SMA Type 2**

Commented on the problem of not having enough funding to maintain the level of care needed and on the availability of carers.

7. Relatives of children / adults with SMA – Total 9

Four grandparents and five aunts/uncles of children responded.

They are related to three children in total, two of whom have SMA Type 3, one of whom has SMA Type 2 / 3. Two of the children are siblings. Replies were therefore about their views of two families who have children with SMA.

Their replies and comments echoed those of the parents.

A representative comment is:

Aunt/Uncle of child age 9 with SMA Type 3

The people of Scotland and my family deserve to have the right to access this medication under the guidance of adequately trained medical practitioners.

I would like to add that hype of this product must always be balanced by a narrative based on reality. Raising hopes and expectations of families without strong foundational evidence is ethically unsound.

Maintaining mobility for as long as possible will be key for both xx and their parents and will allow some freedom and independence without aid whilst going through a critical period into early teenage years.

For my xx if treatment is begun now they might maintain ability to move around the house unaided which will significantly affect the care they will need. The cost and strain of providing care both on my family but also the School and onwards will be immense if the progress of this disease isn't halted. There is currently no other treatment available and clear signs that this treatment will be effective

8. Responses from parents whose child has been treated with nusinersen (including any bereaved) Total 7

Three replies were from parents living in from Scotland; four were from parents living in England

(Gender removed from text as far as possible to protect anonymity)

- **Parent of child with SMA Type 1 / 2, now age 2 years 2 months. Treatment started August 2017 at age 22 months. By 31/12/17 had had 4 doses**

Was only declining but we have now seen improvements since having the drug. More strength in arms, legs and better head control

We hope to see gradual improvements which are ongoing. We hope one day our child can sit independently and eat lumpier foods

For a condition which only worsens this drug is a real hope. To someone who cannot move some improvement is better than nothing. Only families/people in that position can understand what that means. It could mean more years of life

- **Parent of child with SMA Type 1 /2, now age 10 years. Treatment started April 2017 at age 9 years. By 31/12/17 had had 5 doses**

Improved fine motor skills, improved overall movements, improved respiratory functions, improved cough, less chest infections.

Reassurance that something can be done to stop deterioration. Improved respiratory function and cough, improve motor skills and overall movement. Less chest infections.

Nusinersen gave us hope. With less chest infection (which can be life threatening), life is less stressful. It gave us motivation to continue therapies and improve our child's chances to get better. It's wonderful to see improvements for the first time, after so many years of decline in our child's health.

- **Parent of child with SMA Type 1, now age 14 months. Treatment started June 2017 at age 7.5 months. By 31/12/17 had had 5 doses**

My child is now 14 months and can sit unaided for around 30 minutes, can roll from back to side, eats orally, breathes on own with no intervention at all, now uses a stander for 2 hours per day

We as a family are so grateful for nusinersen, we had prepared ourselves for how sma type 1 robs kids of life etc our child has took a whole different path and amazes everyone who sees how well they are doing

My expectations are that my child has the potential to walk, live a relatively normal life

- **Parent of child with SMA Type 1 /2, now age 3 years 8 months. Treatment started February 2017 at age 2 years 10 months. By 31/12/17 had had 6 doses**

It's completely changed our lives. Before nusinersen my child had started the cycle of going in and out of PICU. Every illness resulted in a left lung collapse. We'd always go into hospital with a pretty hefty PICU stay, never the ward. Since starting we've had a couple of very relaxed ward stays as our child needed a small amount of oxygen through their bipap at night. It's been 6 months since our last chest admission and we've handled a few horrific coughs at home, these coughs were far worse than the ones that had our child intubated. They now have a proper cough. If you'd have asked us this time last year how we thought we'd be doing now we probably would have thought we'd lose our baby this year. Now we have a strong, confident amazing child, who is whizzing about in their power chair with far more strength to play. They have head control, regained most of their swallow, can lift some toys, almost sitting, can move legs, can cough, shout, cuddle all the things they couldn't before.

Obviously, a level of worry will always be there, as it's not a cure but we make sure we follow all protocol; we are very strict with bipap, chest physio and general physio which is incredibly important. Nusinersen works alongside protocol it's not instead of. Since starting the treatment and seeing our child's progress, for the first time we've allowed ourselves to look to the future, to think what we'll do in the future, to think about them growing up. Before Nusinersen everything we did was touched with sadness, in the back of our minds we knew that at some point we'd lose our child. Every sniffle or cough was terrifying. It's no life constantly living in fear, wondering if this is the last birthday or Christmas.

There needs to be protocol to go along with Nusinersen, bipap physio etc this will give patients the best chance... it needs to be led by hospitals with amazing respiratory departments.

- **Parent of a child with SMA Type 1 now aged 14 months. Treatment started January 2017 at age 13 weeks 5 days. By 31/12/17 had had 6 doses**

They now have full head control, can sit unassisted for up to 15 minutes with trunk being supported by a lycra suit to help prevent curvature of the spine. Can now move head freely from left to right, and hold in a neutral position. Can easily track people round the room. Can lift arms fully above head when in a slightly reclined seating position and can lift them to shoulder height when in an upright seated position. Can now lift and hold much heavier objects than could ever do in the past. Has increasing hip and leg movement and when lying on back, can lift leg off of the ground and is starting to initiate a roll to the side. With encouragement, can roll right on to side (both sides) however is still reluctant to carry this on further so that is lying on tummy.

Emotionally - We feel that our child is achieving all emotional milestones that would be expected of a typical 14 month old. Practically - can now sit in a supportive chair and play with age appropriate toys, can interact with sibling and peers, sit independently for a short period of time on own and is aware of surroundings and family. Reacts to familiar faces, points to things seen and wanted and is very vocal in babbling.

Physically - At this moment in time we still have to assist with lifting, carrying and transferring a 10kg baby. This is physically demanding and tiring and has resulted in constant back ache and shoulder ache and RSI.

Emotionally - Seeing the amazing progress that our child is making means that we have so much more hope for his future.

With continued nusinersen treatment it is our hope that our child's respiratory system and swallow may strengthen to the point that they have a safe enough swallow for some solid foods to be introduced.

Without the drug nusinersen, the historical nature of this disease means that most babies born with SMA type 1 will most probably die before they reach their second birthday, due to the degeneration of muscles throughout the body but crucially, those around the respiratory and feeding system. Therefore, without the interventional use of this drug, these children will ultimately be condemned to certain premature death.

For those children with SMA Type 2 and 3, I believe the drug nusinersen would absolutely enable them to regain some independence and allow them to regain lost skills or learn to do things that they never could before e.g. a twelve-year-old child who sits daily in a motorised wheelchair, unable to lift their hands beyond their own chest height, could, with nusinersen, be able to lift their hands so that they can then scratch or blow their nose, comb their hair, itch their eye, reach to switch on/off a light switch etc.

The last thing to be considered by the NHS in Scotland is the continued and increasing cost of the palliative care that is required and given to patients all over the country with SMA, versus the cost of a drug that would alleviate some of that burden to the NHS as patients become more mobile and require less physical support. Finally, with other pharmaceutical companies such as Avexis, researching and producing gene therapy drugs which could provide a cure for SMA, I feel that the nusinersen drug will be replaced in time with a complete cure.

- **Parent of child with SMA Type 1, now age 16 months. Treatment started October 2017 at age 13 months. By 31/12/17, had had 4 doses**

Increased movement in legs and arms. Is finding it easier to play and interact with others. Think breathing may have improved also. Still lack of head control. May have caused some vaginal bleeding. Happy as evidence of increased movement. Only ever needed an overnight stay after treatment for monitoring. We have been accepted to have the first of the maintenance doses and we'll be reviewed after this 5th dose.

- **Parent of child with SMA Type 1, now age 2 years 6 months. Treatment started June 2017 at age 2 years. By 31/12/17, had had 5 doses**

Emotionally - has had an impact where our child feels sense of achievement at the improvements they are making, feels more independent & has given a sense of hope that they can do more & more. They feel more included as is able to join in with more toys & games & just in general has more energy so is upbeat & happier.

Practical - able to do simple tasks like feeding self more easily, has more options for play, can now help to wash self & can help with tasks such as baking & helping with practical tasks in the house ... feel more included & part of the family so not missing out on anything.

We hope to see continued improvement & halting of the muscle wasting for our son.

We feel this treatment would be a life saver for people with sma & with the improvements we have seen in our child we feel this treatment should be considered for all types. To see milestones being reached that were never possible before is an incredible achievement for our children. Even to have the ability to sit up, feed yourself, lift & play with toys gives our children so much more opportunity to enjoy life than they would have without this treatment.

9. Responses from those who have been bereaved by SMA - Total 5

	Number
Parents	4
Sibling	1

SMA Type

Type	Number
1	4
2	1

Two respondents said they 'have read a lot about nusinersen'

Representative comments:

- **Parent whose infant had SMA Type 1 and who was bereaved in 2017**

I have seen evidence- based positive progress in all types of SMA. Ultimately treatment must remain a personal decision for each family but I feel the evidence is more than compelling.

I am friends with the parents of two other type 1 families - both children are receiving treatment and started at a young age. Both have had substantial gains in muscle function and increased respiratory health compared to other type 1 children. It is my hope that my child will be the last type 1 baby to die without being offered treatment- every family should have this life-saving option.

Obviously, the gains some families have had are beyond the norm and I do fear newly diagnosed families may have unrealistic expectations. The cost of the treatment is also significant and is a major concern. I feel this will be the biggest stumbling block to allow access for all- but medical professionals should not be putting a price on the lives of children. I would pay anything to get my child back.

Because of the complications in “types” it would not be fair to restrict treatment to just 1. I teach a weak type 2 and I know strong type 1s who outperform weak type 2s on all measures - the gains in respiratory health and muscle function are beneficial to all types. Some gains may seem small to people but even holding a head up independently reduces the risk of choking. Minimal mobility in a hand/arm may mean a young person is able to operate a power chair or type on a keyboard rather than rely on adaptive technology. Any amount of independence is worth its weight. Health care professionals also must consider the potential reduced long-term costs of repeated hospital stays etc.

- **Sibling of a child who had SMA Type 1 and was bereaved in 2000**

This would need to be dealt with on a case by case basis most likely but if a baby was diagnosed with any type of SMA quickly then the treatment should be provided as soon as possible to give that child a proper chance.