NICE National Institute for Health and Care Excellence

Patient organisation submission

Nusinersen for treating spinal muscular atrophy [ID1069]

Thank you for agreeing to give us your organisation's views on this technology and its possible use in the NHS.

You can provide a unique perspective on conditions and their treatment that is not typically available from other sources.

To help you give your views, please use this questionnaire with our guide for patient submissions.

You do not have to answer every question – they are prompts to guide you. The text boxes will expand as you type.

Information on completing this submission

- Please do not embed documents (such as a PDF) in a submission because this may lead to the information being mislaid or make the submission unreadable
- We are committed to meeting the requirements of copyright legislation. If you intend to include **journal articles** in your submission you must have copyright clearance for these articles. We can accept journal articles in NICE Docs.
- Your response should not be longer than 10 pages.

About you	
1.Your name	Liz Ryburn

2. Name of organisation	Spinal Muscular Atrophy Support UK
3. Job title or position	Support Services Manager
4a. Brief description of the organisation (including who funds it). How many members does it have?	We are a charitable organisation that started work 32 years ago providing free information and support to anyone affected by any form of SMA in the UK. We provide a phone, email and home visiting service, and also a 'Shared Experiences' Service. In 2017, we supported 357 adults/families with children living with SMA and are in contact with some 900, including those who are bereaved. We are accredited to the Information Standard and our information sheets are signposted by NHS Choices. Our Research Correspondents have reported on the development of this treatment since trials were initiated. We have contact with clinicians delivering the treatment and had contact with NHS England as it addressed the management of the administrative costs of the SMA Type 1 Expanded Access Programme (EAP), as well as with families wanting access. Our funding comes predominantly from Trusts, the SMA Community and some corporates. This financial year, 2018/19, we received funds from five pharmaceutical companies, including the manufacturers of nusinersen. This was for our core 'outreach' services (6% of overall income) and to cover the costs of our bi-annual information, support and social weekend for families and individuals to be held in April 2018 (9.4% of overall income). We don't receive any government funding.
4b. Do you have any direct or indirect links with, or funding from, the tobacco industry?	No
5. How did you gather information about the experiences of patients and	We invited people to complete our on-line surveys via: a direct email to 605 English households related to people living with SMA / bereaved by SMA; our, other SMA charities and the campaign group TreatSMA's social media channels. We received:

carers to include in your submission?	• 128 returns describing **the health-related impacts of SMA** for 128 people living with SMA Types 1-3. 61% from the adult / young person, 51% from the main unpaid carer. 52% were about people with SMA age 0 – 17 years; 48% about those age 18+ years. • returns describing **the health-related impacts of SMA** for 3 people living with SMA Type 4 • 5 returns from people bereaved by SMA We also sought **people's views on the impact of SMA on their day to day lives and the treatment nusinersen**. We heard from: 56 with the condition; 55 main unpaid carers; 21 other relatives; 5 bereaved by SMA; 26 parents of children treated with nusinersen. In the same way, we sought **the experiences of parents whose children had been treated by nusinersen**. We received 22 replies and added to this replies from our recent Scottish survey – 4 from England, 3 from Scotland. Based on the prevalence of SMA of 1 – 2 in every 100,000, **we estimate we have gathered the experiences of some 14-28 % of those diagnosed with the condition in England.** The full survey results are provided as **appendices 1 - 10.** The numbers and % referred to in this submission relate to these. The picture of the impact of SMA that they paint is confirmed by our Support Services team and our information sheets for families. We speculated that those with stronger views about wanting to access nusinersen may have been more likely to have responded to our survey. We therefore contacted the convenor of what is called the SMA Support UK/SMA Trust 'Adult Insight Group' who is an adult with Type 2. On our behalf, he asked the 35 members if they would want to access nusinersen if it were available. Group members are adults aged 25 – 55 affected by Type 2 or 3 who are interested in having a voice about SMA / disability-related issues. 19 (54%) replied.

Living with the condition	
6. What is it like to live with the condition? What do carers experience when caring for someone with the condition?	SMA is a complex, neuromuscular condition causing progressive muscular weakness and loss of movement. **Types 1, 2 and 3 are childhood-onset forms.** **Type 1** is the most severe - babies are unable to sit without support. Without intervention, most rarely survive beyond two years of age, usually due to breathing difficulties. Some children with **Type 2** sit independently, others require support. Though life expectancy may be shortened, improvements in care standards mean that the majority can live a long life. Children with **Type 3** can stand and walk, although this becomes more difficult and they need support with this over time. Life expectancy is normal. As Types are not rigid categories and there is too much detailed information to present in this summary, the following descriptions of the impact of the condition describe the overall findings from the survey. Clearly some aspects, such as the impact on mobility, will vary according to the severity of the condition and the person's age. This detail can be seen in the full survey results. Our 128 respondents, affected by Types 1, 1/2 and 2 (62%) and Type 2/3 and 3 (38%), with the person with SMA ranging in age from < 2 years to 66+ years, vividly describe their day to day experiences in many pages of responses to the question 'what are the biggest challenges of living with SMA?' It is impossible to do justice to the time and effort they have taken to tell us, a few representative quotes will have to suffice: "The hardest part of SMA for me is the regression….to watch your child lose his greatly achieved milestone it's heart-breaking, you can't explain to him why he can't do that thing he was doing two months ago." **Age 0-2 years, Type 2, father** "My grandson is unable to walk or stand and can sit only with support. He is susceptible to serious respiratory problems….this leads to frequent emergency admissions to PHDU and PICU for up to 5 weeks at a time - the stress placed both on the child and probably more so on the parents in these dangerous situations is immeasurable." **Age 3-4 years, Type 2, grandparent**

	In terms of mobility, 83% use powered wheelchairs, 68% use manual wheelchairs and 21% use wizzybugs – designed for children age 18 months - 3 years who are unable to walk. "As he gets older and bigger the strain of moving and carrying him means more adaptations are needed in the home and less places are accessible. Joining in at school is becoming more difficult. Not being able to go to friends and family's homes. Needing to be turned in the night. Struggling with weight gain. Watching him become less balanced, not being able to sit unaided. Everything getting weaker." **Age 5-12 years, Type 2, aunt/uncle** "My grandson is now unable to walk unaided and uses a wheelchair all the time. He is also slowly losing the strength in his arms. Until the age of 15 he was at least able to walk albeit slowly so you can imagine how frightening it is for the whole family to see how quickly he is deteriorating. It affects us all emotionally, and my grandson physically and practically. He has days when he just can't come to terms with what is happening to him." **Age 13 – 17 years, Type 3, grandparent** Full support – more than would be expected considering the age of the person - is needed for people to go to the toilet (78%), wash (74%), dress (81%), transfer (80%), eat and drink (31%) and, for those who require this, to prepare meals (75%). Between 10 – 42% of others require some support with these tasks. 66% require night care as they are unable to turn over at night or are, for example, needing night time invasive ventilation (29%). For 64% of these, this care is needed between 3 – 6+ times each night. "I cannot do the simplest things on my own: lift my hand to my face, pick up a cup with water, keep my head upright….I cannot go to meet my friends on my own, I cannot go to their houses (not accessible), I cannot hang out with them without having everything pre-arranged so a carer is present." **Age 13-17 years, Type 2, young person** "My son …has become more isolated, doesn't want his friends to see that he can't hold his head up if it falls forward so avoids putting himself in a position where he might need to ask for help and has slowly been pulling away from going out." **Age 13-17 years, Type 2, mother**

	This support is needed because of people's muscle weakness and the other health impacts of the condition: contractures (84%), pain (62%), scoliosis (60%), fatigue with oral feeding (50%), constipation (45%), bone weakness (41%), breathing difficulties (40%) and other health problems. "Physically, I am unable to do anything for myself as all my muscles are that weak now; I cannot walk, stand, transfer, change position independently, hold a pen to write, cannot move or turn over a piece of paper, send a text, use a cash point, clean my teeth, blow my nose, brush my hair, shake your hand, put make up on, scratch an itch, wipe my bottom, feed myself, hold a cup, cuddle my son….." **Age 46-55, Type 2 / 3 years, adult** 48% have no paid support, 25% have between 1 – 10 hours each 24-hour period and 27% have between 11- 24 hours. Respondents described unpaid support for the 128 people with SMA coming from a range of 146 different people with 75% of respondents receiving support from parents. These unpaid carers have other caring responsibilities as well. 51% care for other children, 32% for ageing relatives. Additionally, 39% of the 146 carers had had to give up work completely due to their caring responsibilities, 25% had dropped to part-time. "I am a qualified professional and would love to return to work full time….I am unable to sleep at night as I have to roll my daughter frequently…..All the hospital appointments, treatments, surgeries, etc take up a lot of our time…..I have to do all of the household chores….while my kids are at school, because as soon as my disabled daughter is home she needs my help with everything (bathing, toileting, physio, getting dressed, doing homework, etc). My able-bodied daughter often feels neglected….and I am constantly torn and feel guilty…..SMA has had a huge negative impact on the whole family in every area of our lives - financial, emotional, marital, personal, self-fulfilment and physical health". **Age 5-12 years, Type 2, mother** All those affected by and living with the condition and their carers are describing in their different ways the emotional impact of the condition – the 'chronic sorrow' associated with their ongoing living loss. Symptoms of **SMA Type 4** begin in adulthood and include mild to moderate muscle weakness in the arms and legs and some difficulty walking. This loss of function and the adaptations people who have, until then, often had no physical limitations, is distressing.

Current treatment of the condition in the NHS	
7. What do patients or carers think of current treatments and care available on the NHS?	**Management interventions**, particularly for infants with Type 1, focus on **correct positioning** and ameliorating **breathing difficulties**. These include: chest physiotherapy; oral suctioning; medication to reduce secretions; cough assist; non-invasive ventilation. This is very time-consuming for parents and can be distressing for both them and their child. **Spinal scoliosis**, with its physical and emotional impact, is often managed initially with a lycra suit, spinal brace or jacket but surgery may be recommended if it is contributing to breathing difficulties, preventing comfortable sitting or the curvature has progressed beyond a certain point. 20% of respondents have / have had spinal orthotics; 35% have spinal rods / spinal fusion (54% of those with Type 1-2 aged 18+ years). **Physiotherapy** helps manage contractures and pain, chest physiotherapy (43%) helps manage breathing difficulties. Interventions, particularly for those with Type 2, to manage choking, swallowing, fatigue with feeding, digestion, constipation and managing weight, may include **tube feeding, gastrostomy, medication** and **dietary management**. A major management tool, however, is vigilance and time on the part of carers. To manage the impact of their condition, the children, young people and adults who responded to the survey are having to use **powered wheelchairs** (83%), **manual wheelchairs** (68%), **wheelchair accessible vehicles** (66%), **specialist beds** (63%), **hoists** (60%), **orthotics** (54%), **specialist seating** (50%), **assisted cough machines** (38%), **nebulisers** (31%) and **assistive technology** (30%) as well as other equipment. They require **adaptations to toilet and bathroom facilities** (73%) as well as **other home adaptations** (69%). “Practically our house is full of medical devices and equipment. If we want to go on a trip overnight there is an assisted cough machine and a nebuliser to take, as well as a sleep aid and maybe a specialised chair. Our ‘normal’ is very different from most peoples’.” **Age 0-2 years, Type 2, father** Many described the frustrations they experience in their efforts to secure the support they need in their day to day lives:

	"Being on a wheelchair referral waiting list for so long. Waiting for possible adaptions to house, ground floor bedroom for son as stairs a hazard. As a parent the emotional stress of watching my son's strength quickly deteriorating is unbearable." **Age 5-12 years, Type 3, mother** For 57%, **the number of health and social care professionals** involved range from 6 - 20. Attending appointments and generally managing to coordinate care and support depends on the complexity of the individual's condition and can be very time consuming. Many of the interventions / equipment to manage the condition are **not funded by the NHS** and, although funding may be secured via other statutory sources, many are invariably secured privately or via charitable funding, creating significant financial pressure on families. For example, for these respondents, the NHS is not funding 50% of their powered wheelchairs, 27% of hoists, 36% of toilet and bathroom adaptations, 52% of other home adaptations. Children under the age of 3 years cannot access NHS funded powered chairs so 71% of families find funding for their 'wizzybugs'.
8. Is there an unmet need for patients with this condition?	This is the first drug treatment for SMA. Despite the management interventions that focus on positioning and breathing difficulties, infants with SMA Type 1 rarely survive longer than 2 years. **Spinal surgery** with 'growth rods' means earlier and potentially more effective surgical procedures than previously, but remains daunting for a young child and, as with any surgery, not without risk. Though it results in significant physical and emotional improvements, ongoing vigilance is needed when transferring. Families, especially those with children with Type 1 or 2, spend a considerable amount of time on **daily exercises** to help with contractures and pain and in an effort to maintain mobility. Many comments referred to the **stress of carers** trying to ensure enough is done. Also, despite these management interventions, the stress of trying to avoid chest infections and frequent life-threatening emergency hospital admissions is always there.

Advantages of the technology	
9. What do patients or carers think are the advantages of the technology?	We heard from 29 parents, 27 of whom have children still being treated, 2 of whom are now bereaved:

Doses/injections	Nos.	%
0-4 'loading' year 1	10	34.5
5 - 7	18	62.0
11+	1	3.5

SMA Type	Nos.	%
Type 1	17	59
Type 1 - 2	11	38
Type 3	1	3

Nine parents did not provide any commentary about the impact of the treatment on their child or their family. In their open comments, the other twenty reported already seeing the following advantages for their child:

Total of 20 respondents making 'open' comments	Nos.	%
Physical / muscle improvements	19	95
Much happier	8	40
Respiratory gains	7	35
General improvement in health	4	20
Increased vocalisation	2	10
Tolerates procedure well	2	10
No physical / muscle improvement	1	5
No respiratory gain	1	5
Improved swallow	1	5
Improved quality of life	1	5

"Before treatment he could not even grasp - now he can use both hands to play with toys… he is beginning to hold his head up and can move his legs a little. He has been managing colds all through winter at home whereas before he was in intensive care on life support for every cold he got. He is a happy boy who can now start to explore his

surroundings, he is also beginning to talk … and can sing and clap." **Type 1, treatment started < 7 months, 5-7 injections**

"She has gained skills whereas before treatment she was just losing skills. She has gained head control, more movement in arms and legs. She is able to roll forward which was something she could never do. It has given us all hope. She has stayed off respiratory support and feeding support." **Type 1 / 2, treatment started age 13 - 24 months, 0-4 injections**

"He doesn't fall/collapse as he did before treatment. He fell at least twice a day & some days multiple times. He can now walk faster/further, his gait has improved & is less waddling….He has improved in other motor functions, he's stronger/has more stamina/doesn't fatigue as he did before. He can cycle on the exercise bike and getting better/faster with every treatment. …He can now independently rise from the floor. Emotionally: He is becoming increasingly able and independent which is positively affecting his attitude to life. He has a thirst for knowledge and life. He is exceptional in all subjects at school. He wants to study law and become a lawyer…" **Type 3, treatment started overseas age 12 years, 5-7 injections (See Appendix 11, 1 minute 35 second film clip of before and after treatment)**

This level of ability and drive is something we see frequently in young adults and adults with SMA. How much more could this potential with its positive economic impact be unleashed for them and today's children with treatment?

In their open comments, the following advantages were reported for the parents/family:

Total of 20 respondents made comments	**Nos.**	**%**
Given hope	13	65
Emotionally positive / happier	8	40
Decrease in care needed	4	20
More inclusive family time	1	5
More relaxed	1	5

"This has completely turned our lives around…We were told to enjoy our time left with our child at point of diagnosis and before treatment had become available which was simply heart-breaking. Life as we knew it stopped. Numb with pain and filled with fear we were unable to work/sleep/deal with normal day to day life. However, now I'm witnessing first-hand the benefits of nusinersen I'm simply filled with hope for my child's future. This has had such a positive turnaround for our family, myself, my husband, siblings, grandparents. I feel like I'm no longer waiting on a ticking time bomb, but now look forward to my child's future." **Type 1, treatment started age 13-24 months, 5-7 injections**

When those who have not had treatment and their relatives were asked what they thought the treatment would bring to them / their relative with SMA, 132 responded. The following felt it would bring these advantages:

Total of 132 respondents	**Nos.**	**%**
Will maintain muscle strength	101	77
Will improve muscle strength	96	73
Will extend the life expectation associated with this type of SMA	63	48

44% of them regard themselves as well informed about the treatment, 16% not well informed and 40% 'know a bit'.

"Anything that can increase muscle strength will be life changing for children with SMA, and potentially life-saving if it keeps the respiratory muscles a bit stronger." **Age 5-12 years, Type 2, parent**

"Even if Nusinersen does not provide the desirable results for all patients, clinicians can learn from the results. it will help to develop better drugs that work on wider across SMA spectrum and improve drug delivery mechanisms." **Age 5 - 12 years, Type 2, father**

"The costs associated with having a disabled child are extremely high. With treatment this would be dramatically reduced. Money would be saved on hospital stays, equipment and care. It could also help the economy as it would stop parents / carers having to take time off work /stop working." **Age 5 - 12 years, Type 2, mother**

Disadvantages of the technology	
10. What do patients or carers think are the disadvantages of the technology?	Though the description of the impact of the treatment for 95% of the 20 parents responding is very positive, one had not yet seen respiratory gain and one bereaved parent reported no physical / muscle improvement and also said: "We experienced great distress as a result of conflicting expectations of the likely impact of the drug set by teams in two hospitals. One told us that the drug would slow or even halt the decline. Whereas the other told us that it could reverse the process which would allow him to reach milestones and that he would sit up and possibly even walk. This gave us hope, joy and relief, but later grief when these milestones failed to materialise." **Type 1, bereaved parent** One parent (5%) commented on the emotional distress caused to their child by the treatment, and two parents (10%) commented on the stress it caused them: "It is stressful attending the treatment because as a parent you do not want to put your child through a painful procedure but I feel the benefits far outweigh this." **Type 1, treatment started age 8 - 12 months, 5-7 injections** When asked what impact they thought nuisnersen would have on them / their relative, 9 of the 132 respondents (7%) felt 'it was unlikely to change the natural course of their condition'. When those who had not been treated / whose relative had not been treated were asked for open comments about any concerns they had heard or read about the treatment, 74 responded as follows:

Open comments from 74 respondents	Nos.	%
No/not really	38	51
Lumbar puncture process / safety / discomfort	13	18
Headaches / nausea	6	8
Price	6	8
Scoliosis / spinal fusion may prevent treatment	5	7

Risk of respiratory issues / chest infection	4	5
Side effects	4	5
Unknown long-term outcomes	4	5
No guarantee it will work	2	3
Frequency of treatment / scheduling	2	3

One mother said:

"A lady with a daughter posts details on Facebook of her child's treatment so this is where I have read most details of how it is administered. The injections into the spine put my son off and frightened him. But if he could take nusinersen orally or through his peg he says he might give it a go but I'm not sure if someone so weak like my child would really benefit. When his quality of life is already good he does not want the interruption of keeping going to hospitals for treatment. Now he is older we only have 3 visits a year.' **Age 5- 12 years, Type 2, Mother**

Our understanding from clinicians and families whose children have been treated is that though a lumbar puncture and the need for ongoing delivery isn't the ideal way for treatment to be administered, it is being done successfully and, for many, the procedure is short and straightforward.

As another mother said:

"Every medical procedure carries risk and I would not put our son through these lightly. However, if this were to improve his respiratory and overall muscle function meaning less PICU admissions then we would grab at the chance." **Age 5 – 15 years, Type 2, Mother**

Despite these concerns, 102 of 119 respondents (86%) said they would want the treatment for themselves as a person with SMA / their relative with SMA.

Of the 19 respondents from the adult insight group, 10 (52.6%) would want treatment, 6 (31.6%) wouldn't and 3 (15.8%) would want to see more evidence first:

	"Spinraza - I'd want some really concrete evidence of significant benefit in adults with type 2 before letting them put a needle in my spine. The risk vs benefit is too high for me with the evidence available at the moment. I'd gladly have more or preserved strength and reduced risk of chest infections, but am not convinced Spinraza can do that for me. Lumbar punctures have a risk attached, and where it needs repeated lumbar punctures this increase the risk." **Adult Insight Group member** That said, none of the respondents would want to deny others who wish for the opportunity to access this treatment and interest in other future possible treatments is high.
Patient population	
11. Are there any groups of patients who might benefit more or less from the technology than others? If so, please describe them and explain why.	**138 of 151 respondents (91%) said that they believed that if it is clinically safe for someone with 5q SMA to be treated with nusinersen, they should be given the opportunity to do so.** Asked if they considered any groups might benefit more from treatment (and able to choose more than one option), between 43 - 59% specified the different Types 1 – 3, 32% said Type 4. In terms of those selecting an age group, 52% suggested age 0 – 35 months with a gradual reduction to 35% for age 26 years +. These results must be treated with caution in view of the mix of ages and types represented by respondents. One of the difficulties with judging which groups might benefit more is the lack of clinical trial evidence of the treatment which is thus far only with children aged 0 – 12 years and Types 1 – 3. The evidence of the success of treatment provided to us by the mother of a young person who is now age 13 years and has Type 3 does though highlight the great potential of the treatment to change lives outside this trial group. Many respondents also referred to the positive outcomes that are being reported via social media – particularly from the USA. Clearly there are concerns from people who have a scoliosis and / or have had spinal surgery. We understand, however, from Biogen's community update (21st February 2018) that work is taking place to try to address these challenges.

Equality	
12. Are there any potential equality issues that should be taken into account when considering this condition and the technology?	Local access is very important. Due to the need for regular treatments and the fragility of some, travelling can be very difficult and unsafe. The experience of the slow and uneven geographical roll-out of the EAP and the initial 'postcode lottery' was devastating for many families. The travelling, and the overnight stays needed for some receiving treatment, impacted significantly on those without the financial means / transport. One child and parent we know had to travel huge distances in an ambulance with the other parent following by car while siblings were cared for by relatives. Older children and adults without wheelchair accessible transport of their own will require assistance.
Other issues	
13. Are there any other issues that you would like the committee to consider?	For infants with Type 1, this clearly meets NICE's criteria as an 'end of life' treatment with supportive clinical evidence of its efficacy. Many are seeing this as a potential 'bridge to a cure' with the possibility of future combination therapies. However, in the light of the experience of the bereaved parent, it is vital that information about possible outcomes is clear and expectations are carefully managed. **Emotional and psychological support** is essential. Nusinersen must be supported by **palliative care**, an active approach to care aiming to support the physical, emotional and practical needs of a child and family with a life-threatening condition. Guided by the **International Standards of Care for SMA**, it includes symptom management and reducing complications of muscle weakness. As one parent whose child is being treated said, "It's not a cure… we follow all protocol; we are very strict with bipap, chest physio and general physio which is incredibly important.. it needs to be led by hospitals with amazing respiratory departments." It also needs to be supported by the **swift provision of equipment and housing adaptations**, particularly for some children with Type 1 who may, at least initially as they grow, need lie flat car seats and larger buggies that are not easy to obtain and to date are not NHS funded. Those that are stronger will need access to mobility aids such as the

	'wizzy bug' and appropriate powerchairs – again rarely funded by the NHS to those under age 3 years, despite most children with SMA Type 2 being quite capable of managing these essential aids that enable them to gain independence, access and join in with the world around them. **Starting and stopping criteria** for treatment have been established **for Type 1**, and are used by Centres delivering treatment via the EAP (NHS England's interim policy updated March 2018). We understand that these criteria are discussed with parents before treatment starts so that there is a shared and agreed understanding. Even with this, it can be difficult for medical teams to manage these difficult discussions. The collegial support of the UK-wide NorthStar clinical group is very helpful. Equally, families seeking to make their case for access or faced with treatment being stopped need appropriate support. With further resourcing it should be practically quite possible to roll out the treatment programme to a wider group of children / young people and monitor outcomes through routine clinic visits. The **SMA REACH project,** which already monitors disease progression for 305 children with Type 2 or 3 and the 66 who are receiving nusinersen treatment via the EAP, is an ideal if not essential tool for gathering this data and for further study of the effectiveness of treatment. **For adults,** we suggest outcome goals clearly tied to meaningful day-to-day tasks, as well as more traditional clinical measuring tools would need to be individually agreed between the person and their clinical team. We don't know how 'ready' adult clinicians are to embrace the delivery of this treatment but we imagine many will be keen to offer it to those that wish to access the opportunity. **We suggest treatment should continue unless there is a measurable deterioration.** **Not everyone wants treatment** and this must be respected. We know families with babies with Type 1 who have decided this is not a path they wish to follow. The clinical trial results are good but, as the bereaved parent quoted above shows, they are not guaranteed. Parents may hear that their child could become 'a strong Type 2' but for some, the thought of their child having a lifetime of lumber puncture treatments and living with uncertainty and potentially a very challenging disability (as described), is not a life they feel is right for them or that they could manage for their child or their family.

	The inability to access nusinersen has created huge emotional distress in the SMA community. We sincerely hope this will change: "Small improvements in muscle strength have a disproportionately huge impact on quality of life. So, going from not being able to pick up a drink to being able to do this, for example, is a really big deal. Anything that can increase muscle strength will be life changing for children with SMA, and potentially life-saving if it keeps the respiratory muscles a bit stronger. Watching some of the younger SMA children in the USA hitting milestones and achieving mobility having been treated with Spinraza is really emotional. I wish we could have done that for our son when he was little." **Age 5-12 years, Type 2, Parent**

Key messages

15. In up to 5 bullet points, please summarise the key messages of your submission:

- Day-to-day management of this progressive condition is physically, emotionally and practically hugely demanding for both the person with SMA and their unpaid carers
- Health and social care costs associated with SMA are very high and often not at a level that is sufficient for the person and their unpaid carer(s) to keep physically and emotionally well, get enough sleep, keep socially connected, manage financially and work / study for the hours they wish
- Nusinersen treatment is leading to life-saving and life-changing results for children with Type 1, but it is vital that information about possible outcomes is clear and expectations are carefully managed
- For those with other types of 5q SMA, the small improvements in muscle strength that nusinersen could bring would have a hugely positive impact on their quality of life and health and independence, with a resultant reduction in health and social care costs
- The option of treatment should be supported by symptom management as outlined in the International Standards of Care for SMA, along with ongoing emotional and psychological support for both those with the condition and their carers, and the swift provision of any equipment, home adaptations and care / support packages that are needed to maintain a good quality of life