NICE National Institute for Health and Care Excellence

Patient organisation submission

Onasemnogene abeparvovec for treating type 1 spinal muscular atrophy [ID1473]

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 and Clare Lucas

2.	**Name of organisation**
	Spinal Muscular Atrophy UK (SMA UK) and Muscular Dystrophy UK (MDUK)
3.	**Job title or position**
	Support Services Manager and Head of Policy and Campaigns
4a.	**Brief description of the organisation (including who funds it). How many members does it have?**
	Spinal Muscular Atrophy UK (SMA UK) In September 2018, the charities SMA Support UK and The SMA Trust merged to form **SMA UK**. **SMA Support UK** (previously the Jennifer Trust) had, since 1985 had as its prime focus the provision of free information and support to anyone affected by any form of SMA in the UK. **The SMA Trust** had been funding research since 2003. We are in touch with some 700 households in the UK with a child, young person or adult living with SMA. We estimate this to be over 60% of the total UK population. We are also in contact with more than 350 families who have been bereaved by SMA – the majority by SMA Type 1. SMA UK is accredited to the Information Standard. Our SMA-related information sheets are signposted by the NHS website. In 2018, SMA UK's SMA Type 1 information pages had 17,840 views. That year in the UK, we supported 257 children, young people and adults with SMA and their families via phone, email and home visits. 32 of these were families in England newly affected by SMA Type 1; 3 were families with a child in England with SMA Type 1 whom we had supported previously. Including these 35, we were in touch with a total of 67 families with a child living with SMA Type 1 – via our monthly enews communications (total 2,530 signed up) which keep people abreast of what's happening with services, access to treatments and research related topics.

	Our Research Correspondents (a clinical and a research doctor) report to the SMA community on the development of all drug treatments and clinical trials. We have regular contact with the SMA REACH UK clinical network – which includes clinicians who administer the nusinersen treatment programme and the clinical trials for onasemnogene abeparvovec. Our funding comes predominantly from Trusts, the SMA Community and some corporates. Last financial year, 2018/19, we received funds from four pharmaceutical companies, including the manufacturer of onasemnogene abeparvovec. This was for our core 'outreach' services (4.22% of overall income) We don't receive any government funding. **Muscular Dystrophy UK** Muscular Dystrophy UK (previously known as the Muscular Dystrophy Campaign) was founded in 1959 and has been leading the fight against muscle-wasting conditions ever since. We bring together individuals, families and professionals from more than 60 rare and very rare progressive muscle-weakening and wasting conditions, affecting around 70,000 children and adults in the UK, including SMA. We have 450 individuals on our database with a personal interest in SMA. We provide information, advice and practical and emotional support along with a network of local groups and an online community so that people living with a muscle-wasting condition can find someone to talk to. Every day counts for people with neuromuscular conditions which is why Muscular Dystrophy UK funds pioneering research for better treatments to improve lives today and transform those of future generations. We also press for better recognition of neuromuscular conditions so that people get the best care and support and access to potential drugs much sooner. Our funding comes from donations, gifts, grants and trusts. We have received funds from 11 pharmaceutical companies, including the manufacturers of nusinersen. These were educational grants and one grant for mitochondrial disease research. The funds equate to 0.1% of our overall income. We don't receive any government funding. We received £7,000 from the manufacturers of onasemnogene abeparvovec for our Translational research conference, which other companies also sponsor.
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 carers to include in your submission?**
	In early 2018, in preparation for our submissions to NICE re: the appraisal of nusinersen treatment, SMA UK invited people in the SMA community to complete our on-line surveys. There were: • 128 returns describing **the health-related impacts of SMA** for 128 people living with SMA Types 1-3. Only two of these were from those whose children were affected by SMA Type 1 • 29 returns describing **the experiences of parents whose children had been treated with nusinersen**. The survey responses were integral to the patient group submissions as part of the evaluation of nusinersen. In July 2019 SMA UK and MDUK jointly conducted a survey asking people within the SMA community for their views on the possibility of the NHS funding onasemnogene abeparvovec (for ease referred to as Zolgensma™ in the survey and from now on in this submission). This was disseminated via the charities' (SMA UK, MDUK and TreatSMA) social media channels and SMA UK's monthly e-news. The questionnaire, information sheet and collation of all the 14 responses are in Appendices 1 – 3. This submission draws on: these surveys; the experience and knowledge of SMA UK Support Services Team as a result of its contact over many years with many families affected by SMA Type 1 and MDUK's Information and Support Team's experience.
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 Type 1 is the most severe form of SMA with symptoms usually beginning between 0 and 6 months. Generally speaking, the earlier the onset of symptoms the more severe the condition. Babies are unable to sit without support and may be described as **'non-sitters'**. It's not possible to predict life expectancy accurately but for most children, without intervention for breathing difficulties, this has previously been estimated as less than two years[1]. Evidence suggests that since the International Standards of Care for SMA introduced more proactive management in 2007, children have been living longer[2].

Each child is affected differently, but in general, babies with SMA Type 1 are:

- bright, alert and responsive; their intelligence isn't affected
- able to smile and frown as their facial muscles aren't severely affected
- often described as 'floppy' babies due to their low muscle tone (hypotonia) and severe muscle weakness
- unable to support or lift their head due to their weak neck muscles
- unable to sit unsupported and have difficulty rolling over
- able to move their hands and fingers but have difficulty lifting their arms and legs

They have:

- breathing muscle weakness, which can cause a weak cry and difficulties with breathing and coughing
- an increased chance of chest infections, which can be life-threatening
- difficulty swallowing their saliva and other secretions, which may make them sound chesty or make them cough
- difficulties feeding and gaining weight
- an increased risk of fluids or food passing into their lungs (aspiration), which can cause choking and, potentially, chest infections or pneumonia which can quickly become life-threatening.

Children receive care and support from a multidisciplinary healthcare team including specialists in:

- hospital or community paediatric
- respiratory care
- physiotherapy
- occupational therapy
- dietetics
- speech and language therapy
- palliative care
- general practice and community health care.

This can feel overwhelming.

Positioning is very important. If an infant is too upright or lies on anything that sags or is curved, their chest may concertina or 'hunch up' which makes it more difficult for them to take deeper breaths. During the day they need to have their position changed every hour or so. This helps to relieve pressure to ensure that their joints don't become stiff and gives them a change of

view. Often their neck muscles are weak, and they may need a small neck roll to steady their neck in a more comfortable position and help with breathing. They may be provided with a collar to help and, if they're experiencing tightening of their muscles (contractures) and discomfort, they may have foot and hand splints. As children have a limited range of comfortable positions, they are at risk of developing pressure sores.

Spine, hips and bones

60-90% of children with SMA Type 1 or 2 develop a scoliosis[2]. Children are monitored for this and if there are signs, they may be provided with a spinal brace to wear during the day to help them to sit and breathe more comfortably. It's common for children to have unstable hips which may affect one hip or both and will need monitoring.

Breathing

Weak breathing muscles are common resulting in 'insufficient' breathing which is a leading cause of health problems. To help their child, parents may have to manage:

- Chest physiotherapy to help with comfort and clearing secretions from their child's chest.
- A suction machine to help remove their child's excess secretions.
- Medications that can break down the secretions (such as glycopyrrolate). These have to be used carefully as too high a dose can dry out the secretions too much, which then makes them harder to remove.
- Pain relief if their child is in pain or distress because of breathlessness
- Antibiotics which need to be prescribed quickly when their child is at risk of, or to treat, a chest infection.
- A mechanical insufflator – exsufflator machine (Cough assist) to help clear the secretions from their child's the lungs.
- Oxygen sometimes
- Non-invasive ventilation (NIV) (BiPAP) to help make their child's breathing easier. The SoC guidelines recommend really proactive use of NIV for all infants with symptoms of 'insufficient' breathing and that they start using it early before signs of breathing problems start.
- Short term invasive ventilation if their child has a medical emergency.
- A small number of children may have a tracheostomy

Feeding, nutrition and swallowing

Due to their muscle weakness, a child with SMA Type 1 may have difficulties with feeding and swallowing. Safe swallowing is one of the most important aspects of their care as children with a weak swallow are at risk of inhaling (aspirating) their feed

which can cause choking and respiratory infections. Children often have a weak suck, and mealtimes take longer. Food may get stuck in their cheeks (pocketing) or they may find it hard to open their mouth due to muscle weakness. Infants will need a Video Fluoroscopic Swallow Study and to be monitored for the common problems of gastroesophageal reflux, constipation and vomiting.

If swallowing becomes unsafe, or if a child isn't gaining enough weight, short-term options may include feeding through a nasogastric (NG) or nasojejunal (NJ) tube. A Gastrostomy (PEG) tube is a longer-term option. A Nissen Fundoplication**,** which helps to reduce any reflux, may be done at the same time. Diet has to be very carefully monitored and managed.

Day and Night Care

SMA can make children very sweaty with flushed faces and hot or cold hands. This can make it difficult to judge if their temperature is safe, creating anxiety for their parents. Thin, loose layers of clothing help maintain a comfortable temperature but changing clothing isn't easy, especially if their child is tired or uncomfortable. Parents need to avoid having to lie their child on their tummy due to breathing difficulties. Care is 24 hour 7 days a week.

Impact on Families

The impact of a diagnosis of early onset SMA Type 1 on families is enormous. It often comes as a shock with parents expressing feelings of disbelief, confusion, anger and sadness. The 24 hour-a-day responsibility of caring for a child with complex medical needs that follows is physically, emotionally and psychologically exhausting: constant re-positioning and care, large amounts of medical equipment – many families having to adjust bedroom and living arrangements, the need for specialist car seats and buggies that aren't funded by the NHS, frequent hospital appointments and planned and emergency admissions, involvement of palliative and hospice care, caring for other children, the chronic grief and potential looming loss of their child. Parents describe sleep deprivation, often one will give up or cut back their paid work, social lives disappear. Those that have other children and caring responsibilities can struggle to keep up. The impact ripples out to siblings, grandparents and other relatives and friends, many of whom will try to help in some way, all of whom are also emotionally impacted. Caring for a child with SMA Type 1 also comes with significant financial implications due to the additional costs of living with a disability but also because family members may need to reduce their hours or stop working in order to meet the care needs of the child.

Parents whose children had, in early 2018, begun treatment with nusinersen and responded to our survey that year reflected:

	Before treatment; **"**he could not even grasp …. he was in intensive care on life support for every cold he got." "We were told to enjoy our time left with our child at point of diagnosis 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."
Current treatment of the condition in the NHS	
7.	**What do patients or carers think of current treatments and care available on the NHS?**
	The November 2017 international Standards of Care for SMA (SoC)[2,3,4] outline the minimum care, assessments and interventions families and adults should expect to find in any neuromuscular centre anywhere. This is the current core standard for the management and care of those with SMA Type 1 in England. They include the interventions outlined above. Parents aim for their child to have the best health and quality of life as possible and agree with clinicians that this proactive management and care is essential and should go hand in hand with any potential treatment. Nusinersen is the only currently possible treatment for SMA Type 1. It seeks to increase the amount of SMN protein needed for someone to have healthy lower motor neurons[5]. Without this, these specialist nerve cells in the spinal cord deteriorate restricting the delivery of signals from the brain to the muscles, making movement difficult. The muscles then waste due to lack of use - muscular atrophy. To maintain the necessary levels, nusinersen needs to be delivered regularly over a person's lifetime by intrathecal injection into the cerebro-spinal fluid. It cannot cross the blood-brain barrier. The Nusinersen Expanded Access Programme (EAP) for infants with SMA Type 1 slowly started to roll out in 2017, closing to new patients in November 2018. During this time the vast majority of those with SMA Type 1 were treated – over 80 across the UK. The treatment will now be provided via the Managed Access Programme (MAA). There will be very few if any children with SMA Type 1 who will not be eligible for this treatment. This combined with the SoC is therefore likely to be the current NHS treatment for children with SMA Type 1. When we conducted our survey in early 2018, children had not been on the EAP for long. Of the 29 parents who responded, nine parents didn't provide any commentary about their views of 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." **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." **Treatment started age 13 - 24 months, 0-4 injections**

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… 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.” **Treatment started age 13-24 months, 5-7 injections**

Our recent survey required respondents to read accurate information about the administration and clinical trial outcomes of nusinersen and Zolgensma™ **and** then, score aspects of each one separately on a scale of 1 (strong disadvantage) to 5 (strong advantage). Results for nusinersen were as follows, showing quite a range of opinion:

	Strong Disadvantage 1		2		3		4		Strong Advantage 5		Total
	%	No.	%	No.	%	No.	%	No.	%	No.	
How it is delivered	21.4	3	21.4	3	35.7	5	14.3	2	7.1	1	14
How often it is delivered	35.7	5	28.6	4	14.3	2	7.1	1	14.3	2	14
The range of cells and tissues it reaches in the body	14.3	2	14.3	2	35.7	5	14.3	2	21.4	3	14
The long-term effects on the patient's genetic make-up	14.3	2	14.3	2	28.6	4	14.3	2	28.6	4	14
The potential risks and how these are managed	0.0	0	7.1	1	50.0	7	35.7	5	7.1	1	14
Based on clinical trial results, possible effect on survival / life expectancy	7.1	1	7.1	1	14.3	2	35.7	5	35.7	5	14
Based on clinical trial results, possible effect on breathing	7.1	1	0.0	0	21.4	3	35.7	5	35.7	5	14
Based on clinical trial results, possible effect on motor milestones	0.0	0	21.4	3	21.4	3	28.6	4	28.6	4	14
What is known about this technology	0.0	0	0.0	0	28.6	4	57.1	8	14.3	2	14

8. **Is there an unmet need for patients with this condition?**

Taken at face value, just 14 respondents interested in giving their views on the potential provision of Zolgensma™ by the NHS doesn't show a great unmet need. However, our survey took place at a time when many in the SMA community were completely focused on campaigning and advocacy for the provision of nusinersen for **all** with SMA, which is perhaps why the response rate was so low. In addition, some people may have been overwhelmed by the complexity of the questions being asked and not felt confident responding, despite our best efforts to provide the necessary information to aid their response.

	The scope for Zolgensma™ is for SMA Type 1 only. It would be fair to say as well that any parent with a child with SMA Type 1 awaiting access to nusinersen, and most parents caring for children on the programme already, would be unlikely to have the time, energy or wish to complete a survey that may cause them to question the treatment that had /would imminently be available to them and the road on which they were embarked. Even with the MAA making nusinersen available for those with SMA Type 1, if this new 'one off' treatment offers equal or better potential for quality of life, parents and clinicians need to have the choice to access it. The unmet need is for parents, in consultation with clinicians, to choose what they jointly consider will offer the best potential outcomes for their child.
Advantages of the Technology	
9.	**What do patients or carers think are the advantages of the technology?**
	The Science SMN protein is not just found in the spinal cord, it's also present in all cells as soon as an egg is first fertilised. This means that other organs and parts of the body may be affected by a lack of the protein. Scientists investigating animal models of SMA have suggested that reduced SMN protein may have an impact on the brain, nerves, heart and pancreas. However, only a minority of people with SMA have clearly had challenges with other organs and in those who have, it has not been demonstrated that the cause is the SMA. Research is ongoing[3]. This science does suggest that a treatment that can cross the blood-brain barrier and reach more cells may have an advantage. Our survey required respondents to read accurate information about the administration and clinical trial outcomes of nusinersen and zolgensma and then score aspects of Zolgensma™ on a scale of 1 (strong disadvantage) to 5 (strong advantage). Results were that the Zolgensma™ treatment was seen by the majority to have strong advantages. The views expressed seem to be a reasonable reflection of the comments we hear anecdotally from people and are not unexpected:

	Strong Disadvantage 1		2		3		4		Strong Advantage 5		Total
	%	No.	%	No.	%	No.	%	No.	%	No.	
How often it is delivered									100	14	14
Based on clinical trial results, possible effect on breathing							7.14	1	92.9	13	14
The range of cells and tissues it reaches in the body					7.1	1	7.14	1	85.7	12	14
Based on clinical trial results, possible effect on survival / life expectancy							14.3	2	85.7	12	14
Based on clinical trial results, possible effect on motor milestones							21.4	3	78.6	11	14
How it is delivered					7.1	1	21.4	3	71.4	10	14
The long-term effects on the patient's genetic make-up			7.1	1	21.4	3	14.3	2	57.1	8	14
What is known about this technology			7.1	1	28.6	4	35.7	5	28.6	4	14
The potential risks and how these are managed					57.1	8	21.4	3	21.4	3	14

When asked on a scale of 1 (totally unacceptable) to 5 (totally acceptable) for their view of as a treatment for an infant newly diagnosed with SMA Type 1: 13 (93%) said it was totally acceptable; 1 (7%) considered it acceptable.

When asked to choose one treatment only for an infant newly diagnosed with SMA Type 1 who is well enough to receive nusinersen and who is well enough to receive, and has been screened as suitable for, Zolgensma™ treatment, 13 (93%) chose Zolgensma™ – including two parents whose children are currently receiving nusinersen treatment; 1 (7%) chose nusinersen but stated: 'Only because there is more evidence to show it works. If Zolgensma™ had more case studies to relate to I would chose Zolgensma' (**Parent of child with SMA Type 2 age 0 - 4 yrs. never had drug treatment**); no one chose neither and to only opt for management as outlined in the international Standards of Care for SMA and palliative care.

It's possible that people electing to respond to this survey could be more favourably disposed to Zolgensma™ but the summary information they were required to read was neutrally written (Appendix 2).

	It's also important to note that a small number of parents have elected **not** to have nusinersen treatment and a small number may elect **not** to have Zolgensma™ treatment. Neither treatment is a cure and there is no guarantee of the outcome for an individual child. It's vitally important that, though the earlier the treatment the better, parents have as much time as possible during this very distressing time of receiving a diagnosis to have a fully informed confidential discussion with their clinician about the choices open to them, and that this takes into account emotional, spiritual, cultural and personal circumstances. Parents who choose not to have treatment for their child often express that they support the choice for others.
Disadvantages of the technology	
10.	**What do patients or carers think are the disadvantages of the technology?**
	Our survey required respondents to read accurate information about the administration and clinical trial outcomes of nusinersen and Zolgensma™ and then on a scale of 1 – 5 score aspects of Zolgensma™ on a scale of 1 (strong disadvantage) to 5 (strong advantage). Results were that no one felt there were any strong disadvantages. Potential risks and how they are managed was seen by 8 (57%) as neither an advantage nor disadvantage. These results are a reasonable reflection of the comments we hear anecdotally from people and are not unexpected.

	Strong Disadvantage 1		2		3		4		Strong Advantage 5		Total
	%	No.	%	No.	%	No.	%	No.	%	No.	
How often it is delivered									100	14	14
Based on clinical trial results, possible effect on breathing							7.14	1	92.9	13	14
The range of cells and tissues it reaches in the body					7.1	1	7.14	1	85.7	12	14
Based on clinical trial results, possible effect on survival / life expectancy							14.3	2	85.7	12	14
Based on clinical trial results, possible effect on motor milestones							21.4	3	78.6	11	14
How it is delivered					7.1	1	21.4	3	71.4	10	14
The long-term effects on the patient's genetic make-up			7.1	1	21.4	3	14.3	2	57.1	8	14
What is known about this technology			7.1	1	28.6	4	35.7	5	28.6	4	14
The potential risks and how these are managed					57.1	8	21.4	3	21.4	3	14

Not possible for all children with SMA Type 1

The virus that is packaged with the SMN gene to create Zolgensma™ can be found in the environment; so, some people, including people with SMA, will have a natural immunity to the virus which means the Zolgensma™ therapy as currently delivered will not work for them. Any child being considered for treatment with Zolgensma™ always has a screening blood test to see if they have this immunity and would therefore not be eligible for the treatment.

There may be age, weight and health criteria for treatment which would exclude some infants.

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.**
	We understand from clinical evidence that, as with all the treatments being developed, the earlier the treatment the better the potential outcome, including for those who are pre-symptomatic. As such, there is a need to reconsider newborn screening for SMA.
Equality	
12.	**Are there any potential equality issues that should be taken into account when considering this condition and the technology?**
	This treatment is for children who are disabled with a condition that impacts severely on them, any siblings and family members.
Other issues	
13.	**Are there any other issues that you would like the committee to consider?**
	Access to this 'one-off' intravenous treatment leading to improvements in the outcomes listed in the NICE scoping document would be a step-change in the treatment and management of the condition. It was approved for use in the USA in May 2019. It is important to highlight the one-off aspect of this treatment when conducting the appraisal and considering the budget impact. With clinical guidance, families living with SMA Type 1 should be able to choose the treatment that they consider has the most advantages and the best potential health outcomes for their child.

	We urge the NICE appraisal committee to recommend that newborn screening for SMA in the UK is reviewed by the National Screening Committee as a matter of urgency given the advent of both this treatment and nusinersen, both of which can evidence that the earlier the treatment the better the potential outcome.
	References 1. Wang CH, Finkel RS, Bertini ES, Schroth M, Simonds A, Wong B, Aloysius A, Morrison L, Main M, Crawford TO, Trela A; Participants of the International Conference on SMA Standard of Care (2007) Consensus statement for standard of care in spinal muscular atrophy. J Child Neurol 22: 1027-1049. 2. Mercuri E, Finkel RS, Muntoni F, Wirth B, Montes J, Main M, Mazzone ES, Vitale M, Snyder B, Quijano-Roy S, Bertini E, Davis RH, Meyer OH, Simonds AK, Schroth MK, Graham RJ, Kirschner J, Iannaccone ST, Crawford TO, Woods S, Qian Y, Sejersen T; SMA Care Group. Diagnosis and management of spinal muscular atrophy: Part 1: recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscul Disord. 2018 Feb;28(2):103-115. doi:10.1016/j.nmd.2017.11.005. Epub 2017 Nov 23. 3. Finkel RS, Mercuri E, Meyer OH, Simonds AK, Schroth MK, Graham RJ, Kirschner J, Iannaccone ST, Crawford TO, Woods S, Muntoni F, Wirth B, Montes J, Main M, Mazzone ES, Vitale M, Snyder B, Quijano-Roy S, Bertini E, Davis RH, Qian Y,Sejersen T; SMA Care group. Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. Neuromuscul Disord. 2018 Mar;28(3):197-207. doi: 10.1016/j.nmd.2017.11.004. Epub 2017 Nov 23. **4.** The Guide to the 2017 International Standards of Care for SMA www.treat-nmd.org/care-overview/2017-standards-of-care-for-spinal-muscular-atrophy-sma/the-guide-to-the-2017-international-standards-of-care-for-sma/ 5. ‘More detail on how nusinersen works in SMA.’ www.smauk.org.uk/more-detail-on-how-nusinersen-works-in-sma

Key Messages	
15. In up to 5 bullet points, please summarise the key messages of your submission	
	• SMA Type 1 is a complex and severely disabling condition. Zolgensma™ treatment seeks to address the recent evidence that SMN protein is not just found in the spinal cord, it's also present in all cells as soon as an egg is first fertilised. This means that other organs and parts of the body may be affected by a lack of the protein. • Current treatment will be management as outlined in the international Standards of Care for SMA along with nusinersen. Treatment has to be regular over a child's lifetime with delivery into the cerebro-spinal fluid via lumber puncture. The treatment does not cross the blood-brain barrier • Zolgensma™ is a 'one off' treatment. Delivered via intravenous injection, it can cross the blood-brain barrier • A child with immunity to the virus used in the therapy would not be eligible for the treatment, there may also be criteria of weight, age and health status limiting eligibility • The NHS should fund this ground-breaking treatment which has so much potential. Neither nusinersen nor Zolgensma™ treatment is a cure and there is no guarantee of the outcome for an individual child. It's vitally important that, though the earlier the treatment the better, parents have as much time as possible during this very distressing time of receiving a diagnosis to have a fully informed confidential discussion with their clinician about the choices open to them, and that this takes into account emotional, spiritual, cultural and personal circumstances. Some parents may elect for their child not to have either treatment.

Thank you for your time.

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