Scottish Medicines Consortium
Advising on new medicines for Scotland
www.scottishmedicines.org.uk

NHS SCOTLAND

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Patient Group Submission Form

The Scottish Medicines Consortium (SMC) is committed to working in partnership with patient groups to capture patient and carer experiences, and use them to inform decision-making.

Before you make a submission

You are required to complete a Patient Group Partner Registration Form before you make a submission. The registration form requests general information about your organisation. It only needs to be completed once (and annually updated) and should save you time with any further submissions to SMC. If you have not already completed a registration form, please do this before you make your submission.

You will find it helpful to read our Guide for Patient Group Partners, which gives details about the type of information you need to capture in the submission form. **Please read this before you make your submission and use it to help you complete each question.**

You can find the Registration Form and Guide for Patient Group Partners here:

https://www.scottishmedicines.org.uk/Public_Involvement/Submission_form_and_guidance

Contact us

If you have any more questions after reading the guide, the SMC Public Involvement Team can support you throughout the submission process. You can email us at: hcis.SMCPublicInvolvement@nhs.net or phone: **0141 414 2403.** Please do not hesitate to get in touch, as we are here to help you.

Name of medicine:

Nusinersen (Spinraza)

Indication: (what the medicine is used for)

Treatment of 5q spinal muscular atrophy Types 1, 2 and 3

Submission date:

2nd February 2018

Name of organisation making submission:

Spinal Muscular Atrophy Support UK

Who is the main contact for submissions to SMC?

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Summary of Key Points

Please summarise the key points of your submission which you would like to emphasise to SMC Committee – bullet points may be helpful.

(See P11 of A Guide for Patient Group Partners)

300 words maximum

Quality of life impact / biggest challenges:

- SMA is a complex, progressive, neuromuscular condition
- Without treatment, most infants with Type 1 rarely survive beyond two years of age
- Type 2 and 3 impact significantly on the child/young person/adult's health and well-being, ability to live independently and inclusion in society
- Managing SMA is physically, emotionally and practically demanding for both the person with the condition and their unpaid carer(s)
- Health and social care costs (involvement of many specialists, planned and emergency hospital stays, equipment, adaptations, care packages) are very high
- Delays in obtaining services and caps on statutory funding for equipment and adaptations create frustration and financial pressures for both those with the condition and their unpaid carers
- Health and social care packages and other interventions are 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 the hours they wish.

Limitations of current treatments:

- Nusinersen is the first treatment for SMA
- Current interventions such as those addressing scoliosis, respiratory care and physiotherapy only manage some symptoms of the condition
- No current intervention can arrest the progressive muscle weakness SMA causes.

Benefits of new treatment:

- Clinical trial evidence is that treatment provides the first potential opportunity for children with SMA Type 1, 2 or 3 (age 0 – 12 years) to:
  - achieve physical milestones they would not have otherwise reached
  - maintain physical milestones that they would not have done
  - survive longer than expected considering the typical course of their condition
- Slowing down progression / gaining strength would have a huge impact on independence and reduce costs of health and social care
- Treatment would impact significantly on the emotional and physical well-being of people with SMA and the unpaid carers who support them.

Please provide details of any individuals who have had a significant role in preparing your submission and who have an interest to declare.

(See P11 of A Guide for Patient Group Partners)

No individuals involved in this submission have an interest to declare. 300 words maximum

Please tell us how you gathered information about the experiences of patients and carers to help inform your submission.

(See P11 of A Guide for Patient Group Partners)

300 words maximum

We set up surveys on our website and invited people to complete them via a mailing, our social media channels, other SMA charities and the campaign group TreatSMA.

The prevalence of SMA is 1 – 2 in every 100,000 equating to 55-110 children / young people / adults living with SMA in Scotland.

We received: 29 returns from households related to 12 children with 5q SMA (2 of whom are siblings) and 7 adults with 5q SMA, none of whom had had treatment. We heard from 3 Scottish parents whose children are receiving treatment. We also heard from 5 households bereaved by 5q SMA.

Approximately 60 children have received this treatment in the UK. Using social media, we invited any of their UK parents to complete a survey. We had 4 respondents.

The full survey results are confirmed by our Support Services team who provide a UK-wide phone, email, and home visiting service and from our 'Shared Experiences' Service. (In 2017 we supported 360 UK families and are in contact with some 900 UK households with someone living with SMA.) The results are also backed up by the information sheets we produce for families. These are peer-reviewed by medical experts, cover all types of SMA and include one for families about the

nusinersen Type 1 Expanded Access Programme (EAP). They draw on the experiences of those affected by SMA and our 32 years of working in this area.

Our Clinical and Scientific Research Correspondents have reported on the development of this treatment since trials were initiated. We have followed results. 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 EAP, as well as with families wanting access.

1. How does this condition affect the day-to-day lives of people living with it? (See P11 of A Guide for Patient Group Partners)

500 words maximum

SMA is a complex, neuromuscular condition causing progressive muscular weakness and loss of movement.

Types 1, 2 and 3 are childhood-onset forms of SMA.

Type 1 is the most severe - babies are unable to sit without support. Without intervention, most rarely survive beyond two years of age.

Some children with Type 2 sit independently, others require support. Usually supportive aids for standing and a wheelchair are needed. 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.

Types are not rigid categories.

Our respondents, representing a mixture of those affected by Types 1, 2 or 3 describe a range of impacts including: contractures, pain, bone fractures, scoliosis, acute breathing difficulties, choking, constipation and other health problems.

An adult (24) with Type 2 described the impact of SMA as: “Developing scoliosis meant pain, inability to breathe easily, eat properly and go to school without being in pain.”

Over time, practical interventions to assist daily living include: toilet and bathroom adaptations; wheelchair accessible vehicles; specialist beds; hoists and many other adaptations. For some, there are often long delays in securing these and, as statutory funds often don’t meet full costs, fundraising and applications for charitable help are required.

The parent of a 16-year-old with Type 2 said: “Our teenager’s 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.”

For children, full help or ‘a lot more help than for someone of their age’ is needed for washing, dressing, toileting, transferring, eating / drinking. Night care is needed for three quarters of respondents, with parents providing almost all unpaid care. Paid care packages to help parents and families for children range from 0 to 40 hours / week and for adults, from 0 to 70 – 90 hours / week. Finding and coordinating good paid carers is extremely challenging.

Our survey respondents reported that providing the unpaid care could result in the carer having to give up full time work completely or reduce their working hours, creating financial pressures. 58% of the main unpaid carers are also caring for other children and ageing parents. The level of paid

support is often not enough for them to keep physically and emotionally well, or to get enough sleep and they struggle to keep socially connected. 40 – 60% of adults with SMA felt the same was true for them.

The parent of a child with Type 2/3 SMA said: "SMA 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."

2. How well do medicines which are currently available in NHSScotland help patients manage this condition? (See P12 of A Guide for Patient Group Partners)

500 words maximum

Management interventions, particularly for infants with Type 1, focus on correct positioning and breathing difficulties. This includes: 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. Despite these interventions, infants rarely survive longer than 2 years.

Infants with Type 1/2, or those described as 'weaker' Type 2, also require these time- consuming interventions, often also at night-time.

The parent of a 2-year-old said: "Our child needs turning 4 times a night and the ventilation monitored up to 10 times a night. We have no paid care hours and no relatives to help. Every day is a challenge….we cannot even imagine her frustration, but I can tell you being sleep deprived every single day has been particularly horrible."

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. 53% of respondents have / have had spinal orthotics; 26% have spinal rods / spinal fusion.

A 17-year-old found: "The Lycra suit was a nightmare to get on as so tight. Spinal brace was very hot and uncomfortable to wear."

Another parent of a 9-year-old said: "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."

Spinal surgery with 'growth rods' means fewer 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.

Physiotherapy helps manage contractures and pain. 68% of respondents receive physiotherapy but 16% are awaiting access. Many comments referred to not having enough. One parent of a 2-year-old said: "Physiotherapy is extremely limited….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."

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.

Interventions 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.

The number of health and social care professionals involved with each person can be as many as 7 for adults and 10 for children. Most children require hospital appointments (2 – 6 / month). Attending these and generally managing to coordinate care and support depends on the complexity of the individual's condition and can be very time consuming (2 – 80 hours / month).

3. Have you been able to consult with patients who have used this medicine? (See P12 of A Guide for Patient Group Partners)

Yes ☒ **No** ☐

4. Would this medicine be expected to improve the patient's quality of life and experience of care, and if so, how?

(See P12 of A Guide for Patient Group Partners)

500 words maximum

Biogen's clinical trials evidence of the benefits is compelling as are the survey responses from parents whose children are being treated. All report positive gains.

The parent of a child now aged 14 months with Type 1 who started treatment in January 2017 (aged 13 weeks 5 days) and has had 6 doses said: "…our child is achieving all emotional milestones that would be expected of a typical 14-month-old. Can now sit in a supportive chair and play with age appropriate toys and interact with sibling and peers. Is aware of surroundings and family and reacts to familiar faces. Points to things seen and wanted and is very vocal in babbling."

A child now aged 14 months with Type 1, started treatment in June 2017 (aged 7.5 months). After 5 doses, their parent reported: "Can sit unaided for around 30 minutes, roll from back to side, eats orally, breathes on own with no intervention…now uses a stander for 2 hours/day."

Another parent said their child has: "Increased movement in legs and arms….finding it easier to play and interact with others. Think breathing may have improved… still lack of head control." Their child, now aged 16 months with Type 1, started treatment in October 2017 (aged 13 months) and has had 4 doses.

The parent of a further child now aged 2 years 6 months with Type 1, who started treatment in June 2017 (aged 2 years) and has had 5 doses said: "Our child feels sense of achievement at the improvements they are making…more independent & has given a sense of hope that can do more & more…able to join in with more toys & games… in general has more energy ….able to do simple tasks like feeding self more easily, help to wash self & help with tasks such as baking … feels more included & part of the family."

Another child is now aged 3 years 8 months with Type 1/2, started treatment in February 2017 (aged 2 years 10 months) and has had 6 doses. Their parent reported: "Now we have a strong, confident amazing child… whizzing about in their powerchair with far more strength to play….has head control, regained most of their swallow, can lift some toys, almost sitting, can move legs, cough, shout, cuddle all things they couldn't before."

And finally, the parents of a child now aged 10 years, with Type 1/2 who started treatment in April 2016 (aged 9 years) and has had 5 doses, have seen: “Improved fine motor skills, overall movements, respiratory function and cough, less chest infections.”

Half who have not had access to nusinersen, but feel they know a lot about it, expect that it will slow down or stop the condition’s progress; others expect it would improve strength or recovery of skills, but almost all saying this also said that slowing or stopping of progression would be a worthwhile outcome in itself. 84% of respondents wanted the opportunity for treatment for themselves as adults / their child.

Many regard nusinersen as a bridge to a longer-term potential cure for future children.

5. What kind of impact would treating a patient with this medicine have on the patient’s family or carers? (See P13 of A Guide for Patient Group Partners)

500 words maximum

All parents of treated children reported positively on the impact of the treatment.

One parent of a 10-year-old said: “Nusinersen gave us hope….it's wonderful to see improvements for the first time, after so many years of decline.”

The parent of a child aged 3 years 8 months said: “It’s completely changed our lives….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 about our child growing up. Before Nusinersen everything we did was touched with sadness…It’s no life constantly living in fear, wondering if this is the last birthday or Christmas.”

Another parent put it simply: “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.”

Although not a cure, the potential to slow down or stop progress would have an immense impact on both those with the condition and their unpaid carers - emotionally, physically and practically – reducing dependency and freeing up time that might allow more sleep, less social isolation and more opportunities to live, work and enjoy leisure time in ways they choose. This was powerfully covered by respondents:

The relative of a 9-year-old with Type 3 said: “Treatment begun now might maintain their 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".

One parent stated: "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."

Another said: "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".

A 2-year-old's parent described: "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 their and our lives."

Finally, the impact of untreated SMA and the future people want to avoid was expressed by many, including this 44-year-old who, given their discussion with their consultant about the potential risks due to their scoliosis, commented that they may not pursue this treatment: "Muscle weakness and the reliance it brings on other people is the biggest challenge and frustration. 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."

6. Are there any disadvantages of the new medicine compared to current standard treatments? (See P13 of A Guide for Patient Group Partners)

500 words maximum

This is the first treatment for SMA.

Two 'untreated' respondents expressed some concern at the site of application (lumbar puncture) and one that the longer-term effects were unknown. One adult mentioned the increased chance of respiratory infection and potential liver problems, but added: "But it won't stop me [choosing to access treatment]." One parent of a child having treatment referred to having been advised of the possibility of headaches due to the lumbar puncture, but commented they had avoided this through positioning and extra hydration.

If the medical team and individual / family agree that treatment is proving beneficial, it needs to continue for the person's lifetime and must be regular. In the first year, there are 4 injections in the first 2 months, and a further 2 injections in the remaining 10 months. From the second year, onwards it is anticipated 3 lumbar punctures will be required each year. Though not mentioned by survey respondents as a concern or disadvantage, this requires commitment.

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 a treatment to be administered, it is being done successfully and, for many, the procedure is short and straightforward. We understand that some older children, however, are more anxious and this can be more difficult,

though not impossible, to manage. We are also aware from clinicians that there are potential risks and challenges when someone has scoliosis or has had spinal surgery.

7. Is there any additional information you think may be useful for the SMC committee to consider? (Optional)

500 words maximum

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, especially given that we know from one family, that did not respond to the survey, that treatment doesn't always lead to the positive outcomes hoped for. Guided by the International Standards of Care for SMA, it includes symptom management and reducing complications of muscle weakness.

As one parent said about nusinersen: "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."

Starting and stopping criteria for treatment should be clearly outlined in information sheets with discussion of both with those considering treatment. Medical teams managing these difficult discussions need support, as do families seeking to make their case for access.

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. We know adults with the condition who would not want treatment and question what the search for a cure says about how they are valued by society, but would not wish to deny others the opportunity.

Finally, a parent who tried their utmost to obtain treatment for their child with Type 1 who died in June 2017 summarises:

"I am friends with parents of two type 1 families - both children started treatment 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 fear newly diagnosed families may have unrealistic expectations. The cost of treatment is also significant and a major concern… - but I would pay anything to get my child back.

The gains in respiratory health and muscle function are beneficial to all types. Some gains may seem small 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."

Top-line survey results, % of 'untreated' respondents

- Adults with, or parents of child with, Type 1/2 or 2 (57.5%)
- Adults with, or parents of child with Type 2/3 or 3 (42.5%)

Reported health impacts:

- contractures (63%)
- scoliosis (63%)
- constipation (58%)
- pain (53%)
- bone fractures (37%)
- acute breathing difficulties (37%)
- choking (36%)

Aids and adaptations required:

- home toilet and bathroom adaptations (84%)
- specialist beds (68%)
- wheelchair accessible vehicles (63%)
- hoists (58%)

Care for young children:

- dressing (79%)
- toileting (79%)
- washing (77%)
- eating / drinking (63%)
- transferring (58%)

8. Do you consent for a summary of your submission to be included in the Detailed Advice Document for this medicine?

Yes ☒ No ☐

Thank you for completing this form.

Please email it to: hcis.SMCPublicInvolvement@nhs.net

If you are unable to email this form to us, please send by post to the address below:

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The Public Involvement Team is available to advise you on how to complete this form to ensure the patient and carer experience is fully captured, to help inform the SMC decision making process. If you have any questions about completing this form call us on: 0141 414 2403.