Healthcare Improvement Scotland | SMC Advice on new medicines

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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* in the Public involvement and Making a submission sections of our website.

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: his.smcpublicinvolvement@nhs.scot

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)

“For treatment of 5q spinal muscular atrophy (SMA).

Submission date:

04/07/2025

Name of organisation making submission:

Spinal Muscular Atrophy UK (SMAUK) and Muscular Dystophy UK (MDUK)

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

Nusinersen (Spinraza) is a clinically proven, disease-modifying treatment that offers significant benefits to individuals with spinal muscular atrophy (SMA) types 2 and 3. It slows disease progression, stabilises or improves motor function, and enhances quality of life.

1. Proven Clinical and Quality of Life Benefits

- Patients across Scotland and the wider UK report improved strength, mobility, respiratory function, and independence.

- For many, these gains reduce their care needs, improve daily functioning, and positively impact mental wellbeing and family life.

- Regular hospital visits for nusinersen provide structured clinical monitoring and reassurance for patients and carers.

2. Patient and Carer Testimonies

- Families describe it as “life-changing,” giving hope and reducing psychological strain.

- Stability of condition through nusinersen enables patients to remain independent, maintain education or work, and reduce pressure on carers.

- Carers report improved mental health and reduced anxiety about the future.

3. Treatment Preference and Patient Choice

- Some patients prefer nusinersen’s less frequent administration (three times a year) over daily oral alternatives like risdiplam.

- A treatment choice between nusinersen and risdiplam ensures individual needs and circumstances are respected.

- Uncertainties around risdiplam’s impact on male fertility means it is not the first choice for some young men.

4. Access Inequities in Scotland

- Adults in Scotland cannot currently receive nusinersen locally.
- Lack of an adult treatment centre has skewed real-world data and denies fair access.
- Children doing well on nusinersen face being forced off the treatment upon transition to adult services, creating distress and instability.

5. Importance of Fair and Equitable Access

- Restricting access by SMA type (e.g. only Type 1) is clinically inappropriate due to overlap in disease severity and outdated classifications.
- Without accounting for SMN2 gene copies or diagnosis history, some patients may be wrongly excluded.
- Scotland's lack of procedural flexibility (e.g. options other than general anaesthetic for children) further limits access.

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 Partner*s)

300 words maximum

No declarations to make

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

-Surveys

In 2024, we set up surveys on our website and invited people to complete them via a mailing, our social media channels, MDUK and the campaign group Treat SMA.

We received:

26 returns from people prescribed nusinersen and living with type 2 and 3 SMA in the UK, including 2 from Scotland.

55 returns from carers of people prescribed nusinersen living with type 2 and 3 SMA in the UK, including 3 from Scotland.

In 2025 we distributed a survey for patients and their carers living in Scotland. We received 11 responses, 4 living with SMA and 7 parent carers.

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

- Wider knowledge, understanding and lived experience of the SMA UK team

Our Support Services team provide a UK-wide phone, email, and home visiting service. They draw on the experiences of those affected by SMA and our 40 years of working in this area.

Since 2023 the SMA UK team have moderated 5 community Whats App groups where we listen and learn from the SMA community as well as advising on key issues raised.

Within the staff team, 3 are parents of children living with SMA (one living in Scotland), one is a sibling of an adult living with SMA type 2 and one is an adult living with SMA type 3. All have had lived experience of nusinersen treatment.

SMA UK and MD UK are members of the SMA REACH UK steering committee and work closely with specialist SMA clinicians across the country.

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

Type 1 is the most severe; without intervention babies cannot sit unaided and most do not survive past two years. Children with Type 2 may sit independently or with support but usually require mobility aids. While life expectancy can be shortened, improved care now enables many to live longer. Type 3 children can stand and walk initially, though this ability diminishes over time. Life expectancy is generally normal.

SMA exists on a spectrum, and the classification depends on age of symptom onset, severity, and clinical judgment. The role of SMN2 copy number in classification has only emerged since disease-modifying treatments became available.

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

One respondent aged over 40 with progressed SMA type 2 SMA reflected how SMA impacted her life and the lives of her children before and after nusinersen treatment;

‘ There is hope and there is evidence via the assessments that I am not deteriorating. I can cut up my own food now, eat more than soft food only, brush my hair now, I feel like a person again and not simply a burden. I have children and want to be there for them, this drug gives us all the life we want. Before Spinraza I was losing strength and stamina and my children were missing out in comparison to other children.’

Across the severity spectrum, SMA has a significant impact on the ability to carry out day to day tasks. A young adult respondent living with SMA type 3 reflected on the impact of SMA and the differences she had experienced with nusinersen treatment;

‘My strength has improved tenfold. My core is stronger which means I can go out and about in my chair more often, lean forward to brush my teeth over the sink, and cough easier; my arms are stronger, making it easier to dress, wash, dry myself, make tea or a snack, pick up books for university, and play piano. These strength improvements have also greatly improved the quality of my life in all aspects.’

Without treatment, those living with type 2 and 3 SMA, will, over time, require practical interventions to assist daily living including: toilet and bathroom adaptations; wheelchair accessible vehicles; specialist beds; hoists etc. There can be long delays in securing these and, as statutory funds often don’t meet full costs, fundraising and charitable help are required. Paid support is often insufficient, affecting carers’ sleep, emotional wellbeing, and social life.

100% of carers surveyed reported that SMA caregiving had a significant impact on their physical and mental health. They cited fatigue, stress, fear for the future, back pain, reduced income, and strained relationships. Practical burdens included managing care, arranging appointments, and the high costs of holidays, housing adaptations, and mobility aids.

One parent of a child with Type 2 SMA shared:

‘ I’ve had to leave my job as a result of poor health which comes from frequent night get ups and physical pain from lifting. I get depressed thinking about what my son can’t and won’t be able to achieve.’

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

Adults and children living with SMA types 2 and 3 in Scotland can currently access risdiplam, a small molecule drug that targets the ‘back up’ survival motor neuron 2 (SMN2) gene to produce more SMN protein, thus prolonging the survival of the motor neurons.

Risdiplam is an oral liquid medication that must be taken once a day. Once prescribed, a free home delivery service is set up, enabling patients to manage the treatment in their own home.

Listening to the community through 1:1 support, social networking groups, face-to-face workshops and wider community surveys, SMA UK can surmise that risdiplam does not impact everyone in the same way and treatment outcomes are varied. As with all the disease modifying treatments for SMA, the paediatric population, treated earlier in disease progression, sees the most significant gains. Risdiplam has also been transformative for the type 2 and 3 adult population, bringing disease stability and small but hugely impactful motor gains that enable people to maintain the level of independence and the quality of life that they had adjusted to before treatment.

‘I have more energy and can better control my head and neck when driving outside in my wheelchair - this means I can go out without a carer, and even this small gain in strength has given me a huge amount more independence and freedom. I'm a university student, and since taking Risdiplam I've been able to walk around campus with friends, because I've not been worried/needed support for my head control. Before this gain I found it virtually impossible to go anywhere without a carer, and now that I can I've been able to meet new people and make new friends, simply by being able to go from one place to another without help. This small gain has had an immeasurable impact on my life’ 20-30yr old living with SMA type 2 in Scotland, risdiplam patient.

There are also reports of individualised gains in other areas, such as energy levels and bulbar function.

However, there are those who have not responded to Risdiplam as well, one person experienced increased fatigue, another had ‘debilitating’ gastric side effects. There is no clinical evidence that these experiences are attributable to Risdiplam. However mild gastric symptoms have been reported internationally.

Due to a lack of data in human studies, and linked to data from animal studies, the risdiplam label warns of the unknowns around the impact of risdiplam on fertility, this impacts treatment decision making, particularly young men.

The only other treatment option for those living with type 2 and 3 SMA in Scotland is best supportive care. Management interventions, (particularly for type 2’s at the more severe end of the spectrum), focus on correct positioning and breathing difficulties. This includes:chest physiotherapy; oral suctioning; medication to reduce secretions; cough assist; non-invasive ventilation and a team of clinicians working together to manage the multisystemic impact of the disease.

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

Nusinersen patients across the UK report both improvement and/or stability across a wide range of functions including strength, mobility, fine motor skills, respiratory health, and bulbar function. Through SMA UK's surveys, our virtual networks, face-to-face community events and our own personal experiences, the message is clear that these physical gains, or stability in function, has had a direct impact on quality of life and mental wellbeing.

'My child can now walk independently, she is strong and follows everyday patterns compared to her peers unaffected by SMA. Because of treatment, our quality of life is not impacted by my daughter's SMA' Parent carer of child living with SMA type 2 in Scotland nusinersen patient aged 5

For adults living with a progressed form of the disease, stability has brought an enormous sense of relief and hope.

'It's been a real mental soother to know I'm not getting weaker in theory and my results prove it.'

We have heard from adult Spinraza patients and their clinicians that they value the regular clinic visits. Having to go to hospital for treatment three times a year ensures that they are regularly monitored, with opportunities for formal assessments that clinically validate their stability or gains in function.

In our most recent survey to the SMA community in Scotland, 3 out of the 7 respondents living with type 2 or 3 SMA were currently nusinersen patients, and 4 are risdiplam patients.

Given the limitations to risdiplam listed above, and the fact that responses to treatment are individualised, it is imperative that adults living with SMA type 2 and 3 right across the UK have a choice of the two available disease modifying therapies. This is the only way that people can work hand in hand with their clinicians to find the best fit for them individually and thus the best treatment outcomes.

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

Family and carers of people living with SMA, unanimously report how nusinersen has improved their day-to-day life, in particular their psychological wellbeing. The impact on family and carers generally increases with the level of SMA severity.

'Looking after our son with type 2 (weak) has changed our lives, the constant alert you are on knowing that any illness or cold could have a very damaging effect. He has been on Spinraza for 4 years, it's changed all our lives and lessened the burden on our mental health and anxiety for the future.'

The impact on social life and relationships within the family unit was also reported, with parents able to focus their time on each other or siblings.

'I feel if my daughter was not on any treatment, fear, anxiety would over take me and I wouldn't be able to enjoy socialising as much.'

'My husband and I have no time alone together. It is an extraordinary juggling act to be able to spend time with the other 2 children 1 on 1. It's hard'

A significant impact that living with SMA has on family financial resources was also commented on by 100% of the 2024 survey respondents.

‘The cost of living with a person with a disability is extreme. Funding house adaptions, holidays, day trips, transport; the list is endless. Also trying to work to provide income to fund day to day living is hard when I have to take time off to care for my daughter.’

A lack of reliability in the access to treatment also causes anxiety. There are many nusinersen patients whose trust in the efficacy of the treatment means they would not want to switch to a daily oral medication, the prospect of being forced to switch treatment induces much anxiety for some.

‘We see everyday my son happy living independently we support him but he is just living his best life. The treatment has given us, as a family, hope and we can see our son grow up to have a fulfilling life. It's been a life line for us as a family and we have remained strong watching him get stronger. I don't think it would be the same outcome if we didn't have this treatment. We would be absolutely heart broken seeing him lose anymore than he already did.’ Parent of child living with SMA type 2 in Scotland, nusinersen patient

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

Nusinersen is delivered intrathecally at a specialist centre. This is an invasive procedure that requires a short hospital admission 3 times per year. The suitability of this treatment pathway varies depending on individual preference:

‘The experience has been and always has been positive, we take 90 minutes to get to the treatment centre. My son has been looked after incredibly well since 2020. The whole care team around him are incredible human beings from theatre staff to his consultant nurses, Dr's, play therapists to the lovely HCAs they have all been amazing and looked after my son so well. The treatment takes around 45-60 mins in theatre then we stay 5 hours in total then home with no issues every time’.

The experience could be improved further for Scottish patients if there was more flexibility in the management of the procedure, as highlighted by a parent of a child living with type 1 SMA in England;

‘After 45 minutes waiting for the numbing cream to take effect, the procedure takes about 10 minutes. We are then observed for an hour and are free to go home soon afterwards’

Spinal fusion for treatment of scoliosis makes an intrathecal procedure more difficult, yet not impossible. Two out of the four risdiplam patients who responded to our Scottish 2025 survey, made their treatment decision because spinal fusion restricted their access to nusinersen.

The other two adult respondents, one living with type 2 and the other with type 3 reported that risdiplam was the only treatment choice offered to them given that there is no service set up for the intrathecal procedure in Scotland. This is a situation replicated in adult care across Scotland and has had a hugely significant impact on the ability of the orphan drugs programme to collect any real-world data on the efficacy of Spinraza on adults living with SMA in Scotland.

7. Are there any potential equality issues that should be taken into account when considering this condition and medicine? (See P13 of *A Guide for Patient Group Partners*)

500 words maximum

Although nusinersen has been available via the ultra- orphan drugs programme in principle to those living with type 2 and 3 SMA in Scotland, there are significant issues with access which have restricted access to both paediatric and adult populations.

-No Scottish nusinersen treatment centre for adults

Currently, if an adult living with SMA in Scotland was to choose nusinersen treatment, they would have to travel to Newcastle for the intrathecal procedure. With no clinical centre in Scotland set up to deliver nusinersen, children living with all types of SMA who are currently thriving with nusinersen treatment will have limited choices when transitioning to adulthood. Being forced to switch from a treatment that is working well will cause undue stress for the whole family.

-Restricting access to type 1 SMA only

Expanding access to nusinersen to type 2 and 3 SMA in Scotland would reduce the inequities that enabling access only to those disgnosed with type 1 would create. As stated above, and highlighted in fig 1, the symptoms of SMA sit on a severity spectrum – those living with a severe form of type 2 SMA, could well have been clinically classified as type 1 if they had had a different diagnosing clinician. Those diagnosed before the introduction of SMA treatments will not have had their SMN2 copy number confirmed, and so have an even higher chance of sitting within an inaccurate type class. Limiting access to types is not a fair or equitable system.

-No alternative option to General Anaesthetic administration for children in Scotland

One parent of 7 year old twins living with SMA type 2 in Scotland found that despite the obvious efficacy of Spinraza, the procedural anxiety of general anaesthetic, and the side effects from the anaesthetic became too difficult to manage;

'Both of our children were on Spinraza for just over 4 years however we switched to risdiplam due to a number of factors primarily medicine being given under GA (general aneasthetic).'

Unlike the flexible procedural pathways followed in England, an alternative sedation or numbing cream was not offered as an option.

-No newborn screening for SMA in the UK

Newborn screening for SMA is the only way to see the best health outcomes from treatments, and the most equitable outcomes. Current evidence shows that newborn screening would significantly reduce many these challenges in the future and completely irradicate some of them.

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

500 words maximum

Treatment for SMA is not a cure, to get the best health outcomes from nusinersen treatment, it must be supported by best supportive care. This involves input from a multi-disciplianary team, assessing and managing the symptoms of SMA.

With the current standards of care out of date, we, as partner patient organiisations, are working with the REACH clinical network to rewrite the standards of care in SMA through the project SMA Care UK. This project will produce reccomendations for care and management in each discipline based on an initial individualised assessment.

9. 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.

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, please email it to:
his.smcpublicinvolvement@nhs.scot