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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](https://www.scottishmedicines.org.uk/about-us/public-involvement/) and [Making a submission](https://www.scottishmedicines.org.uk/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.scotPlease do not hesitate to get in touch, as we are here to help you.Name of medicine:

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| Risdiplam |

## Indication: (what the medicine is used for)

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| Treatment of 5qSMA |

## Submission date:

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| 4th October 2021 |

## Name of organisation making submission:

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| Spinal Muscular Atrophy UK and Muscular Dystrophy 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*)

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| • SMA is a complex and often severely disabling condition300 words maximum• For people impacted by SMA, a progressive muscle wasting condition, the importance of stabilisation and even the smallest benefit of a treatment cannot be stressed enough.• Risdiplam offers a daily home-based oral treatment. 97% of the SMA community we have consulted (2020) consider home delivery to be either a strong advantage or advantage.• The treatment crosses the blood brain barrier. Five of six people treated via the risdiplam EAMS who responded to our 2021 Scottish survey describe experiencing reduced fatigue and improved fine motor skills. Four also described improved swallow, breathing and increased strength. The one other person stated it was too early to say. • In our 2020 survey of 137 respondents 16% had negative / very negative views about the potential impact on male fertility and 10% on female menstruation (10%). Our follow up questions to Roche suggest this can be managed.• All our Scottish survey respondents wish to see NHS Scotland funding risdiplam. Some parents whose children currently receive nusinersen would swap treatment.• We hope the SMC will recommend that, irrespective of age, all who have 5q SMA will have the opportunity to have NHS funded access to risdiplam treatment, with a decision to go ahead or not based on a grounded and realistic discussion with their clinician about the potential benefits and any risks to them individually. |

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)

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| 300 words maximumLiz Ryburn Support Team Manager SMA UKKate Adcock Director of Reaesrch and Innovations MDUKNo interests to declare |

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*)

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| 300 words maximumThe SMA community has been frequently surveyed concerning the development of treatments and their clinical and cost effectiveness. We therefore chose not to run a further survey about the impact of SMA and drew instead on the 128 responses to the joint charities’ (SMA UK, MDUK, TreatSMA) 2018 surveys about the impact of SMA. These included 29 responses from people in Scotland, with an age range of people with SMA between < 2 – 65+ years: https://smauk.org.uk/our-surveys-about-the-impact-of-sma-and-views-about-access-to-nusinersenOur first UK-wide survey on whether people affected by SMA would wish risdiplam treatment to be funded was conducted 25th Sept - 18th Oct 2020 with 137 responses (adults with SMA - 52%; parents of young people < 18 years old with SMA - 23%). The clinical classification of the person with SMA described in the survey response was: Type 1 (7%); Type 2 (50%); Type 3 (38%):https://smauk.org.uk/sma-uks-and-mduks-joint-submission-to-nice-risdiplam SMA UK and MDUK’s Scottish SMA community survey on this topic ran 2nd – 16th September 2021. It was emailed directly to 55 households where there is an adult or child who has SMA known to SMA UK. It was also promoted by both patient groups on social media. There were 29 responses relating to 23 children / adults who have SMA describing their treatment / non treatment experiences and views of risdiplam. Our SMA population estimate for Scotland based on a prevalence of 1 – 2 in 100,000 is between 55 – 110, with a mid-point estimate of 82 – a response rate of 28%. Our submission is also informed by SMA UK’s Support & Outreach Service contact with many adults, families and community networks.  |

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

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| SMA’s severity varies between and within ‘Types.’ From the 2018 survey: More than 80% used powered wheelchairs and / or manual wheelchairs (70%) and almost a quarter of children aged 1.5 – 3 years used powered ‘Wizzybugs’ “….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.” 0-2 years, Type 2 (Parent) “He is unable to walk or stand and can sit only with support. He is susceptible to serious respiratory problems… frequent emergency admissions … up to 5 weeks at a time - the stress….is immeasurable.” 3-4 years, Type 2 (Grandparent) “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…...He has days when he just can't come to terms with what is happening to him.” 13-17 years, Type 3 (Grandparent) Full assistance to simply transfer from one position to another, including to the toilet, and to dress was needed by 80%, while 75% needed this to wash and almost a third to eat and drink. Three quarters needing to prepare meals relied on others. Lack of independence has a major impact on many people’s dignity, especially older children and adults.Dependence often continues at night, with two thirds unable to turn themselves and needing a carer / PA 3 – 6+ times a night, and almost a third needing non-invasive ventilation (NIV).“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…. go to meet my friends on my own, go to their houses (not accessible), hang out with them without having everything pre-arranged, so a carer is present.” 13-17 years, Type 2SMA creates many health challenges. Contractures (80%) causing pain (62%) and loss of movement; scoliosis (60%), uncomfortable orthotics and invasive surgery. Half reported oral feeding fatigue, constipation and breathing difficulties. Bone weakness (41%) also creates many risks.“Physically, I am unable to do anything for myself… I cannot walk, stand, transfer, change position independently, hold a pen to write… 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…” 46-55 years, Type 2/3Almost half had no paid support, and for others, it ranged from 1- 24 hours. Parents cared for 75% of the 128 who relied on 146 unpaid carers. 39% of unpaid carers had stopped work, 25% went part-time; 51% cared for other children, 32% for ageing relatives. “I am a qualified professional and would love to return to work full-time…I cannot sleep at night as I have to roll my daughter frequently…. the hospital appointments, treatments, surgeries…. she needs my help with everything (bathing, toileting, physio, getting dressed, doing homework, etc). My able-bodied daughter often feels neglected …SMA has had a huge negative impact on the whole family… financial, emotional, marital, personal, self-fulfilment and physical health.” 5-12 years, Type 2 (Parent)Demonstrating the spectrum of ages of individuals with SMA, one of our survey respondents said:"I am a wheelchair user, with no ability to stand or walk, and the only thing I can do with my hands is move and click a computer mouse. The daily assistance that my wife now provides includes: dressing, washing me and teeth cleaning, feeding, toileting, and all other tasks requiring the use of hands. Whilst a child, I suffered humiliation. Now aged 71, I experience guilt about being the cause of the burdens on her.""So, I achieved some things in my life. But I was not physically able to hold my three new-born children in my arms or hold my three new-born grandchildren or go out for a walk with any of them. I could not do anything physically to care for them then or in their childhoods and I cannot do anything physically now to help my wife, whose arthritis is making her caring increasingly difficult and painful. I don't want to finally lose the last remaining functions of my hands. It would be especially good to recover sufficient strength to lift some of the demands made of my wife/carer." 71 years, Type 3All, in different ways, described the emotional impact– the ‘chronic sorrow’ associated with ongoing loss.  |

1. 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

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| When considering this question, we ask people who have SMA what aspects of their condition they would most want to manage.When medicines for SMA were first assessed for efficacy, the emphasis was on motor milestone gains and survival. For young children, there is now more understanding of the importance of treatments on respiratory outcomes, and other outcomes that impact on quality of life, such as the development of: swallowing, feeding / eating; verbal communication; muscle support of spine and hips; fine motor skills and fatigue levels.For older children, young people and adults, there is much greater awareness of the impact the condition has on upper limb movement and the related importance of fine motor skills and fatigue levels which enable or maintain independence and improve quality of life as described above (Q1). Importantly there is an understanding that this is a progressive muscle wasting condition. In 2019, 96.7% of 1,327 validated responses to SMA Europe’s survey of people diagnosed with SMA stated they would “consider it to be progress if there was a drug to stabilize their current clinical state.” Though it’s important to remember people will rate outcomes based on their current ability levels, it may still be helpful to note what the 137 respondents in our 2020 survey some 77-84% considered increased independence, improved stamina and reduced fatigue, improved fine motor milestones and improved motor milestone to be very important outcomes if they could be impacted by a treatment. These outcomes would also reduce dependence on carers.Nusinersen treatment for many children has resulted in positive motor milestone, fine motor skills and respiratory outcomes. However, it does not cross the blood-brain barrier as it is delivered intrathecally via lumbar puncture every four months for life, which can be a challenging experience for a child. As yet, there is no delivery in Scotland of this treatment to adults for whom this is clinically suitable. Additionally, for the majority of those who have severe scoliosis / spinal surgery, it is challenging to administer and is therefore not considered clinically suitable.The one-time gene therapy zolgensma has shown promising results and is now funded by NHS Scotland for infants who have SMA Type 1 who meet clinical and safety eligibility criteria. The treatment may not be possible for a very small percentage of infants who have SMA Type 1. It is not available for older / heavier children or for any child or adult diagnosed with SMA Types 2 or 3.From September 2020 until July 1st, 2021, risdiplam was potentially available via an Early Access to Medicines Scheme (EAMS) for those who have SMA Type 1 or 2 for whom nusinersen was considered to be clinically unsuitable. The scheme was never an option for anyone who has SMA Type 3. Now the EAMS scheme has closed, going forward there is no treatment option for: any child for whom nusinersen is considered to be clinically unsuitable; or any adult, whether or not nusinersen were a clinically suitable option.  |

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

Yes [x]  No [ ]

1. 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*)

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| 500 words maximumClinical trial results have demonstrated positive motor milestone and fine motor outcomes and, importantly, meaningful improvements for people as shown via the patient-reported SMA independence scale.The 137 respondents to our 2020 survey were invited to read risdiplam information and clinical trial summaries compiled by SMA UK and MDUK Research staff. Respondents considered the following aspects of the treatment would be a strong advantage / advantage that would impact on their quality of life: How taken - syrup by mouth (96%); How often – daily (75%); How long for (71%); Where (home) (97%); Where stored - fridge (71%) “It’s non-invasive and can be self-administered at home without medical professionals. That’s a milestone.” Adult241 people in the UK are on the risdiplam EAMS (23.5% of the 1,025 mid-point estimate of people with SMA in the UK). There are many positive reports of experiences.In Scotland,12 adults and 3 children take risdiplam via the EAMS and one child via the Compassionate Use Programme (total 16 - 19.5% of the midpoint estimated Scottish population with SMA). Our 2021 survey had full responses from 37.5% of these, all with SMA Type 2: 4 adults - one via their parent, and 1 young person and 1 child – via their parents. Asked about the impact the treatment has had on them, two said it was too early to say. The other four all described reduced fatigue, improved fine motor skills and strength. Three also described improved swallow and breathing. All wish to continue.‘I felt a change within days. I can move arms with more ease to my mouth. Not as tired as I used to get. Go out much more on a daily basis whereas I would be tired if out one day. Gaining more strength & movement with the help of daily exercises. For the first time in over 5 years I blow dried my own hair, couldn`t believe it. When I go out for meals, I have had to use a tray on my wheelchair but on a few occasions when I can get into the tables properly, I have managed without it’. 55 – 64 years Type 2‘After three months I feel a little bit stronger every day from day one and also I sleep so much better after taking Risdiplam. I have stronger neck, stronger arms and legs, I can seat longer, and I am not tired at all. I can brush my teeth again by myself’. 25 – 34 years Type 2Risdiplam crosses the blood brain barrier creating greater potential for reaching more cells in the body; nusinersen, cannot cross the blood brain barrier. One parent noted in their survey response that this was their key reason for stating that if they had the option, and it was clinically suitable for their child, they would swap to risdiplam.At a public meeting, we heard a leading clinician commenting on her early observations that this treatment seemed to be having a positive impact on both swallowing and respiratory function. |

1. 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*)

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| 500 words maximumThe comments from our survey respondents illustrate the impact the treatment has had on the person who has SMA and, by implication, the impact this will have had on caring responsibilities. Added to this there is a clearly positive emotional impact for carers.‘My daughter has been taking Risdiplam since December 1st, 2020, the difference we have noticed have been amazing what may not seem big differences to a healthy person as a parent of an SMA daughter the differences are huge. Risdiplam has changed my daughter’s life she was diagnosed in 2003 so we waited 19 years for a treatment to watch her lift her hand to apply her mascara, going through a day without napping to holding a pint glass with her juice in it and so much more has been unbelievable to watch. She is also now learning to drive, and I believe without Risdiplam she would not have managed to accomplish this.’ 19 – 24 years, Type 2 (Parent)‘Almost every day now I cannot stop and sit to do nothing, I just feel full of energy and have been doing a lot more. Doing crafts/painting. Eating is less of a struggle. This has given me a new lease of life. And for my family it has been quite overwhelming’. 55 – 64 years, Type 2In our September 2020 survey, this response vividly captured the positive impact for carers due to this being an oral treatment delivered at home: “The lack of requirement to have a surgical procedure with risk of infection is a plus. Loss of school days for visiting hospital 150 miles return in a day is of enormous benefit. At a time when hospital visits are only possible in emergency cases home treatment and administration is a definite positive to reduce risk of catching COVID 19 on journeys and in hospital. It releases clinicians to do other essential work.’ Family memberHome delivery is especially important for more rural families, especially those whose travel can be so affected by inclement weather. It means less time off work for carers and more flexibility for families in terms of, for example, being able to take holidays.This treatment takes away any stress around the ongoing four-monthly lumbar puncture treatments – stress which can increase with children as they get older. In our September 2021 survey 4 of the 8 respondents whose 5 children are currently receiving nusinersen said that if they had the opportunity and it was clinically suitable for their child, they would swap to risdiplam citing this as their main reason:‘Risdiplam would be easier to administer as it’s oral. Spinraza is great but requires anaesthetic and being in hospital’ 0 – 5 years Type 1 (Parent)‘Because I don’t want my children to have a general anaesthetic and lumbar puncture their whole life’ 0 – 5 years Type 1 (Parent) |

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

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|  500 words maximumIn our September 2021 survey, three of the five taking risdiplam reported no side effects. One said it was too early to say. The two others commented:‘After five days, I was a little tired. A slight pain in the muscles in the arms. I had this only in the first month’. 25 -34 years, Type 2‘A little upset stomach for the first couple of weeks but nothing else negative’ 19 – 24 years, Type 2 (Parent)In our 2020 Survey, we asked the 137 respondents for their views on aspects of the treatment that were known so far. There were negative / very negative views about the potential impact on male fertility (16%) and on female menstruation (10%). We have followed up these concerns with the company who state that there is no cumulative impact on male fertility for boys and that in adults the impact is reversible. We are aware that some men have chosen the option of sperm banking as a precaution. In terms of the impact on women’s fertility, no SMA treatment trials are undertaken with pregnant women in this cohort. Roche advise that no woman trying to conceive or pregnant should take the treatment and that this would be the advice for many treatments.  |

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

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| 500 words maximumAll 2021 survey respondents (31) wish risdiplam to be funded by NHS Scotland:Of the ten adults not having treatment, seven would want risdiplam; others didn’t answer this question. In our survey, of the eight people taking risdiplam, seven (who responded to all questions) wish to continue, one didn’t answer this question. Four of the eight parents of children on nusinersen/spinraza would wish to swap to risdiplam. The other four stated they ‘don’t know’:‘As it’s never been available to us we’ve never fully investigated the results. An oral intake of medicine would be much better than intrathecal injection under anaesthesia every 4 months, if the outcomes are comparable’ 0 - 5 years, Type 1 (Parent)‘I haven’t thought about it with my child doing so well on spinraza’ 0 - 5 years, Type 1 (Parent)‘I don’t know if risdiplam would be better than spinraza’ 0 - 5 years, Type 1 (Parent)There are no head-to-head trials comparing treatment outcomes.‘The clinical classifications and ‘Typing’ of SMA was introduced in 1990 by a committee of clinicians and geneticists to promote collaborative studies between different centres and to identify the genes of SMA. Their classification was based primarily on the age of onset and the age of death, with the ability to sit unaided and stand and walk unaided added on. The classifications were never meant as a way to make decisions about who should / should not have access to treatment’ (V Dubowitz writing in ‘SMA Disease Mechanisms and Therapy’ edited by Summer, Paushkin & Ko, 2016). We’re aware ‘adult onset’ Type 4, isn’t included in this application, however, we note all ‘Types’ are part of the 5qSMA form of SMA which is a continuum. Young adults may ‘deny’ symptom onset, have symptoms dismissed or find the road to diagnosis delayed. One man we know, diagnosed with SMA Type 4, looks back and can see that his symptoms started before he was 19 years. Numbers classified as Type 4 are very small, life expectancy is normal; a treatment that could stabilise or improve progressive muscle weakness would greatly improve quality of life.JEWELFISH had an upper age limit of 60 years. We suggest this was about integrity of trial data and shouldn’t govern potential access. One respondent aged 71 has SMA Type 3 and is keen for treatment.There appear to be people within the patient population in Scotland with an SMA diagnosis who are unaware of the new treatment possibilities; in some cases healthcare professionals are also unaware. As a patient group, while recognising this is not the responsibility of the SMC, we consider improving awareness of SMA treatments and compiling a national database of people with SMA to monitor progress in the delivery of treatment to be an essential developmnent .There are a number of Scottish Government strategies for the NHS in Scotland and for unpaid carers. The strategic objectives of these include 'to ensure the people of Scotland experience the best quality health and care services' and 'to maximise the benefits from national policies and approaches intended to support carers'. Provision of this new treatment would open up extraordinary opportunities for children and adults with SMA and their family carers. This would be a major achievement in the delivery of these strategies. We’re concerned about the potential for geographical inequalities in accessing treatment. Many adults and children use powerchairs and, with the support of personal assistants and / or parents / carers, manage a complex and challenging disability due to their progressive muscle weakness. Travel is always logistically challenging and though treatment is home-based, initial health assessments need to be clinic-based. It is therefore important for treatment to be delivered as close as possible to where people live. |

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

 Yes [x]  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