



**UK National Screening Committee
Screening for spinal muscular atrophy
Consultation comments pro-forma**

Name:	Liz Ryburn	Email address:	office@smasupportuk.org.uk
Organisation (if appropriate):	Spinal Muscular Atrophy Support UK and The SMA Trust (merging to form Spinal Muscular Atrophy UK)		
Role:	Support Services Manager		
<p>Do you consent to your name being published on the UK NSC website alongside your response?</p> <p align="center">Yes <input type="checkbox"/> yes No <input type="checkbox"/></p>			
Section and / or page number	Text or issue to which comments relate	Comment	
		<i>Please use a new row for each comment and add extra rows as required.</i>	
Page 7.	Criterion 1, the prevalence of SMA in the UK has not been met following the review update	<p>We refer to the following studies which were also reviewed by the NSC:</p> <ul style="list-style-type: none"> • Verhaart I <i>et al.</i> (2017) Prevalence, incidence and carrier frequency of 5q–linked spinal muscular atrophy –a literature review. <i>Orphanet J Rare Dis</i> 12: 124. • Verhaart I <i>et al.</i> (2017) A multi-source approach to determine SMA incidence and research ready population. <i>J Neurol</i> 264: 1465-1473 	

		<p>These conclude:</p> <p>Prevalence - between 1 and 2 people in every 100,000 worldwide have a type of 5q SMA. In 2017, the UK population was approximately 66 million. Based on this, it is estimated that between 660 and 1320 people have SMA in the UK at any one time.</p> <p>We are aware that these papers are based on global observations of incidence and prevalence, but until we have an accurate UK-wide register of those born with 5q SMA, and those already living with 5q SMA, we suggest these give the most accurate available data.</p> <p>We note that the NSC is concerned to identify the prevalence of all forms of SMA, irrespective of genetic aetiology. However, the NSC's criteria 10 focuses on reviewing the effectiveness of pharmacological treatment for SMA. Currently, the only treatment available for SMA (Nusinersen) is exclusively targeted to those with confirmed 5q SMA, which is by far the most common form of SMA. It is therefore the prevalence of 5q SMA that is important when considering whether this condition meets NSC criterion 1.</p> <p>In summary: we suggest that there is enough global evidence of the prevalence of 5q SMA. If the NSC requires further information about the prevalence in the UK, we ask that they work closely with NICE which is addressing the very same question in its deliberations about whether to recommend the treatment nusinersen for NHS funding (see below). However, if other criteria are met, we ask that any current population uncertainties do not hold up the possibility of progressing a 5q SMA newborn screening programme.</p>
Page 7.	Criterion 1 'there is no evidence as to how many	We refer to the above studies, which conclude an incidence of 5q SMA of approximately one in every 10,000 babies worldwide. In the UK in 2017, there were 755,043 live births.

	<p>people are affected by each type of SMA</p>	<p>This suggests that, in that year, approximately 76 babies were born with a Type of 5q SMA. These studies further conclude the incidence of the types of childhood onset SMA:</p> <ul style="list-style-type: none"> • Type 1: 60% infants age < 6 months* – suggesting 46 infants each year • Type 2: 21% children ages 6 – 18 months – suggesting 16 children each year • Type 3: 19% children including - suggesting 14 children each year, including: <ul style="list-style-type: none"> ○ Type 3a ages 18months – 3 years ○ Type 3b age 3 years plus <p>(*we note the NSC review page 11 suggests only 50%)</p> <p>We acknowledge that the % of type is based on global studies and that there is an urgent need for more work to be commissioned to establish this accurately in the UK context. However, the Expanded Access Programme (EAP) providing nusinersen treatment for those with SMA Type 1 has now been nationally available since August 2017, with children’s progress tracked via the SMA REACH project. We understand that some parents of infants diagnosed with SMA Type 1 are declining treatment, but that number is now very small and should be known by treating clinicians. This means we are now very close to having much more accurate data on the incidence of this type of SMA in the UK.</p> <p>We have highlighted to NICE that work to establish population numbers and type of SMA is urgently needed if they are to progress a much-needed Managed Access Agreement for nusinersen. We suggest that in the interim, the percentages above, combined with average life expectancy for those with each type of SMA could be a sufficient guide for numbers.</p>
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		<p>In summary: If other criteria are met, we ask that any current population uncertainties do not hold up the possibility of progressing a 5q SMA newborn screening programme.</p>
Page 7	<p>Criterion 1. It is not yet possible to accurately determine from an individual's genotype whether they will be mildly or severely affected by SMA</p>	<p>The age of onset of symptoms of 5q SMA and the <i>SMN1</i> gene deletion test quickly (2-4 weeks) inform a diagnosis of 5q SMA Type 1, 2, 3 or 4.</p> <p>The <i>SMN2</i> copy number has also been seen as a potential way of further establishing the future course of the condition. However, the internationally agreed Standards of Care for SMA (2017)^{1,2} (SoC) show the narrow variance between the 'usual' number of <i>SMN2</i> copy numbers compared with the possible 'range' described by Tillmann <i>et al</i>³. Those with:</p> <ul style="list-style-type: none"> • Type 1 have a 'usual' <i>SMN2</i> copy number of 2 but a 'range' of 1-3 copies • Type 2 have a 'usual' <i>SMN2</i> copy number of 2 but a 'range' of 2-4 copies • Type 3a have a 'usual' <i>SMN2</i> copy number of 3 but a 'range' of 3-5 copies • Type 3b have a 'usual' <i>SMN2</i> copy number of 4 but a 'range' of 3-5 copies <p>As stated in the 2017 SoC, at the individual level, perfectly accurate predictions cannot be made about the type or severity of SMA based on <i>SMN2</i> copy number alone. This is due to other genetic and possibly environmental factors that have a small influence on the disease course.</p> <p>We note, however, that the information about <i>SMN2</i> copy number and other tests are considered as sufficient to predict the severity of an infant's 5q SMA when used in newborn screening programmes in the USA (see below)</p> <p>We are aware that there is also significant ongoing research into the predicted future disease impact, which includes both genotype and looking at biomarkers.</p>

		<ol style="list-style-type: none"> 1. Mercuri, E <i>et al.</i> (2017) 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. 2. Finkel, R <i>et al.</i> (2017), 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.) 3. Tillmann, A <i>et al.</i> (2018) Spinal Muscular Atrophy (SMA) type 1, a changing phenotype: Implications for motor function and physiotherapy management from the Nusinersen Expanded Access Program (EAP) APCP Journal Volume 9 Number 1)
Page 8	<i>Criterion 4. There should be a simple, safe, precise and validated screening test.</i>	<p>The NSC review states that, ‘it is not possible to robustly quantify the accuracy of screening methods for SMA neonates’ We note that the NSC has evaluated four studies and finds that they provide weak evidence for such a test. Again, these tests are seeking verification of 5q SMA.</p> <p>We note the further study by Kraszewski, J.N <i>et al.</i> (2017) Pilot study of population-based newborn screening for spinal muscular atrophy in New York state Volume 20 Number 6 June 2018 GENETICS in MEDICINE which reports on newborn dried blood spot (DBS) screening of 3,826 newborns screened at three hospitals in New York City from January 2016 to January 2017. They were tested for 5q SMA using the deletion in exon 7 of <i>SMN1</i>. We don’t know if the NSC would regard this study as sufficiently robust given its other critique’s, but note that as a result of this study, one infant was enrolled in the NURTURE clinical trial (see below) and was first treated with Spinraza (see below) at age 15 days. She is now age 12 months, meeting all developmental milestones, and free of any respiratory issues.</p>

		<p>In summary: newborn screening tests are being used to identify infants with 5q SMA. If such testing is introduced it would need to be made clear to parents that the test is for 5q SMA, the most common form of SMA with clear information about the accuracy of the test and that this test does not cover all possible other very rare forms of SMA.</p>
<p>Page 8.</p>	<p><i>Criterion 9. There should be an effective intervention for patients identified through screening, with evidence that intervention at a pre-symptomatic phase leads to better outcomes for the screened individual compared with usual care.</i></p>	<p>As the NSC review points out, the 2007 Consensus statement on the Standards of Care for SMA, subsequently published as the ‘International Standards of Care for SMA’ have focused on management of the condition. Largely due to the improvements in care and management that they have driven, there’s been increasing evidence that people with SMA and their families can expect a better quality of life than in the past. Their recent updating in 2017 capture the many changes there have been in management of the condition and will no doubt be the NSC’s reference base in any further base.</p> <p>Nusinersen treatment</p> <p>As the NSC review points out, Nusinersen, which is marketed as Spinraza™, is the first disease-modifying treatment for SMA. The review considers the results of Biogen’s clinical trials (ENDEAR and CHERISH) showing the positive outcomes of nusinersen treatment in Children with SMA Type 1, 2 and 3. A further trial, SHINE, looking at longer term outcomes is ongoing.</p> <p>The following publications confirm outcomes in ‘real world’ studies of the nusinersen Expanded Access Programme for SMA Type 1¹⁻⁵ All indicate that the earlier the treatment, the greater the benefit:</p>

		<ol style="list-style-type: none"> 1. Europe - 33 children aged from 8.3 to 113.1 months - December 2016 - May 2017. Aragon-Gawinska, K <i>et al.</i> (2018) Nusinersen in spinal muscular atrophy type 1 patients older than 7 months. A cohort study <i>Neurology</i>[®] 2018;00:1-7. doi:10.1212/WNL.0000000000006281 2. Australia – 16 patients aged 2.5 months to 35.7 years November 2016 – September 2017 Farrar, M <i>et al.</i> (2018) Nusinersen for SMA: expanded access programme <i>J Neurol Neurosurg Psychiatry</i> 2018;89:937–942. doi:10.1136/jnnp-2017-317412 3. England - Great Ormond Street Hospital – 21 patients aged 8.3 – 113.1 months March – October 2017 Tillmann, A <i>et al.</i> (2018) Spinal Muscular Atrophy (SMA) type 1, a changing phenotype: Implications for motor function and physiotherapy management from the Nusinersen Expanded Access Program (EAP) <i>APCP Journal</i> Volume 9 Number 1 4. Germany – 61 patients aged 1 – 93 months in seven neuromuscular centres November 2016 – June 2017 Pechmann, A <i>et al.</i> (2018) Evaluation of Children with SMA Type 1 Under Treatment with Nusinersen within the Expanded Access Program in Germany <i>Journal of Neuromuscular Diseases</i> 5 (2018) 135-143 DOI 10.3233/JND-180315 5. Italy – 104 patients – aged 3 months – 19 years 9 months - first six months of EAP Pane, M <i>et al.</i> (2018) Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function <i>Neuromuscular Disorders</i> 28 (2018) 582-585 30 May 2018
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		<p>The NSC review refers to results published by Bertini <i>et al.</i> in 2017 from NURTURE, the ongoing open-label, single-arm study evaluating the efficacy and safety of nusinersen in pre-symptomatic infants with genetically confirmed SMA and comments on the improvements that infants have made, though criticises that the study did not compare pre-symptomatic treatment to treatment after symptoms start. We suggest that this is being addressed through further studies, Biogen’s long term follow up clinical trial, SHINE, of all treated with nusinersen to date and the now many ‘real world studies of treatment that are ongoing.</p> <p>We note Biogen’s March 12th, 2018 summary of the interim Phase 2 results as this model and its outcomes appears to be underpinning the newborn screening programmes that are now operating in the USA:</p> <p>In the NURTURE study, SPINRAZA was administered to infants six weeks old or younger, who were in the pre-symptomatic stage, genetically-diagnosed with SMA and had two or three copies of the <i>SMN2</i> gene (n=15 for two copies (most likely to develop Type 1 SMA); n=10 for three copies (most likely to develop Type 2 SMA)). At the time of this interim analysis, infants had been followed for up to 25.6 months – well beyond the typical timeframe when most infants with Type 1 SMA would have required permanent ventilation or died. The interim analysis, showed that all infants were alive, and none required tracheostomy or permanent ventilation. All showed improvement in motor function and motor milestone achievements as of July 5, 2017, compared to the disease’s natural history.</p> <p>Dr. Darryl C. De Vivo, M.D., lead study author said, “The NURTURE findings document the continuing benefits that SPINRAZA provides for patients with SMA</p>
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		<p>who initiated treatment in early infancy while clinically pre-symptomatic, including age-appropriate developmental gains in motor function and motor milestone achievements,” and “The treated infants in the NURTURE study had genetic SMA and were likely to clinically develop Type 1 or 2, yet with enough observation time they have all achieved independent sitting and the majority have developed the ability to walk.”</p> <p>NURTURE participants also achieved a mean CHOP INTEND score, which measures general motor function among infants with SMA, of 58.4 at last visit (out of a maximum score of 64). Many continued to improve and maintain these scores beyond a point in time at which untreated individuals with Type 1 SMA would experience a significant decline. Overall, the study showed that SPINRAZA was well-tolerated and no new safety concerns were identified.</p> <p>Though still early on in the study, and without the benefit of a full analysis of how much greater the gains are for these children compared with children who have been followed via the ENDEAR study, results seem to suggest greater efficacy of pre-symptomatic treatment and therefore greater potential cost savings : Finkel, R <i>et al.</i> for the ENDEAR Study Group (2017) Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy n engl j med 377;18 nejm.org November 2, 2017 states ‘Approximately half the infants in the nusinersen group who received permanent assisted ventilation did so within 13 weeks after they received the first dose; this result indicates that a minimum treatment time is required to see the full benefits of nusinersen. This result, as well as our finding that infants with a disease duration at screening longer than the median duration of 13.1 weeks were more likely than those with a disease duration no longer than the median duration to need permanent assisted ventilation, suggests that early initiation of treatment may maximize its efficacy.’</p>
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		<p>In summary: there is a growing body of clinical trial and real-world evidence of the effectiveness of nusinersen treatment for 5q SMA and that the earlier the treatment the greater the impact, with strong indications that the greatest impact occurs if it is started pre-symptomatically.</p> <p>Other treatments</p> <p>There are also other treatments on the horizon which are coming close to completion of clinical trials and, one imagines, possible applications for licences (AveXis’ AVXS-101, Roche’s RG7916 / risdiplam).</p> <p>We note that these are also indicating that the earliest possible treatment has the greatest impact.</p>
Page 9	<p><i>Criterion 10: There should be agreed evidence-based policies covering which individuals should be offered interventions and the appropriate intervention to be offered</i></p>	<p>Screening programmes in the USA</p> <p>In terms of screening of SMA neonates, we note that several US states have recently introduced newborn screening. Those with between 1 and 3 <i>SMN2</i> copies are offered nusinersen treatment and those with 4 <i>SMN2</i> copies (carried by the majority of those with SMA Type 3b) are monitored. Newborn screening for 5q SMA is now, we understand, on the Recommended Uniform Screening Panel (RUSP), which makes it much more likely that additional states will adopt it.</p> <p>In summary: Effective screening programmes for 5q SMA are now operating globally which, as well as the confirmed diagnosis, use information gained from <i>SMN2</i> copy numbers to establish which children should be treated and which monitored. These are establishing an evidence-based policy as to which individuals should be offered interventions and the appropriate intervention to be offered.</p>

		<p>Access to treatment in the UK</p> <p>As the NSC knows, nusinersen treatment has been available in the UK for those with SMA Type 1, via Biogen’s global Expanded Access Programme (EAP). Since August 2017, this has been limited to those who are <7 months of age, Biogen has announced that this programme will close on 1st November 2018.</p> <p>On 7th May 2018, Scotland agreed to fund the treatment for those with SMA Type 1. On 18th June 2018, the Scottish Government announced <i>it is introducing a new definition of 'ultra-orphan medicines' that can treat very rare conditions affecting fewer than 1 in 50,000 people - around 100 people or less in Scotland</i>, advising that this would be implemented on 1st October 2018. Biogen has made it known that it has applied for nusinersen to be the first drug to be appraised for those with SMA Type 2 or 3 via this new system.</p> <p>The NSC also refers to NICE’s current appraisal of the treatment. Clinicians, patient and parent groups are working hard to respond to NICE’s initial report, which does not recommend funding. Clinicians, patient and parent groups are pushing for a reversal of this decision and access for all with SMA Type 1,2 or 3 SMA or, at the very least a Managed Access Agreement. There is currently no clinical trial evidence for treatment of those with the much rarer SMA Types 0 or 4.</p> <p>In summary: In Scotland an effective treatment, nusinersen, is available for those with SMA Type 1. We await the possibility of this being extended to those with Type 2 or 3. We also await the outcome of NICE’s decision making as to who will be eligible for treatment in England and Wales. This should be finalised by 21st November 2018. We understand Northern Ireland may follow either Scotland or England.</p>
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		<p>There is growing clinical evidence that a newborn screening programme will be essential for ensuring greatest potential benefit from nusinersen treatment and, in due course other treatments. This screening programme would identify children who will develop SMA Types 1, 2 or 3. which will allow the earliest possible monitoring and best possible management based on the SoC. However, unless treatment is available to all these children, it will lead to unimaginable distress for families. NICE and the Scottish Medicines Consortium and the NSC must coordinate their thinking and planning.</p> <p>In summary: at this stage it is not known which children will be able to access nusinersen treatment in the UK. This decision and the possible criteria for a proposed screening programme are critical to any further debate and decision making</p>
Page 17.	Ethical implications and dilemmas	<p>We note and agree there are considerable ethical implications and dilemmas posed by newborn screening. This includes whether it should be an opt in or opt out programme. Furthermore, current screening programmes for 5q SMA will, we imagine, also potentially identify those who will at some point in their lives develop adult onset Type 4. The current method and life-long delivery required for nusinersen would not be an option. Though screening offers the possibility of monitoring, creating a scenario whereby families has to live with this knowledge creates significant ethical challenges.</p> <p>In summary: any screening programme must be accompanied by a robust and ongoing programme of supportive counselling and accurate information about: what information may be revealed via the screening test; what impact the various types of 5q SMA have on life; what management and treatments are available.</p>
Page 17	SMA Community Views – do people want access to nusinersen?	<p>In January 2018 we conducted a survey about the impact of SMA on children, adults and their families. We also asked for views on access to nusinersen.</p>

The responses of parents/carers to the question ‘Would you want your child to have access to nusinersen?’ is most relevant to this submission. Respondents were aware that the treatment would involve repeated lumbar puncture deliveries over the person’s lifetime and that long-term outcomes were not known.

56 parents/carers responded. 95% said yes, they would want their child to have treatment, and 5% said no.

Of those that said yes, their child had the following SMA Type and is the age shown below:

SMA Type	1	1 / 2	2	2 / 3	3
%	4	60	2	32	2
Age (years)	0-2	3-4	5-12	13-17	18+
%	13	11	34	8	34

The 5% that said ‘no’ have children aged 13 – 17 years who have SMA Type 2.

This does perhaps indicate that, even with the uncertainties of long term outcomes and the method and frequency required for delivery of the treatment, it is likely that the uptake for those with children screened for treatment would be very high.

SMA community views – attitudes to newborn screening

We already know from Dr. Felicity Boardman’s work that the SMA community is largely supportive of newborn screening, particularly when compared to other forms of screening that involve the potential loss of SMA lives.

		<ul style="list-style-type: none">• Boardman F, <i>et al.</i> (2016). Newborn screening for spinal muscular atrophy: the views of affected families and adults. American Journal of Medical Genetics 173A: 1546-1561. <p>Dr Boardman's paper on the views of the general population on newborn screening for SMA indicates that 84% of those surveyed (232) were in support of newborn screening for SMA is also salient:</p> <ul style="list-style-type: none">• Boardman, F <i>et al.</i> (2017). Newborn genetic screening for SMA in the UK: the views of the general population. Molecular Genetics and Genomic Medicine, 1-10. <p>The introduction of nusinersen is likely to only bolster this support as shown by the follow up study we conducted in August 2018 – a survey of community views on newborn screening (survey monkey link sent out via social media and our monthly e-news).</p> <p>As access to nusinersen has been at the top of the agenda for the SMA community this year - focusing on raising awareness with the media and MPs and taking part in NICE's appraisal, it was not surprising that this only elicited 19 responses. However, results were:</p> <ul style="list-style-type: none">• 84% of respondents were affected by SMA Type1; 37% were bereaved by SMA Type 1.• 95% strongly agreed that newborn screening should be introduced in the UK, 5% neither agreed nor disagreed.• 89% strongly agreed and 11% agreed that it should be a programme similar to ones in the States that offer treatment to those with between 1 and 3 <i>SMN2</i> copies and that
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		<p>those with 4 copies of <i>SMN2</i> (carried by the majority of those with SMA Type 3b) should be monitored</p> <ul style="list-style-type: none">• 84% strongly agreed and 16% agreed that a newborn screening programme that led to the earliest possible treatment would, in the long run, result in better quality of life for children and families <p>Comments included:</p> <p>‘My grandsons diagnosis took agonising months as local GPs kept fobbing us off. Clearly, they had no idea what was wrong. Screening at birth would alleviate all those months of anguish and better prepare parents for care and treatment.’</p> <p style="text-align: right;">Bereaved Grandparent of child with SMA Type 1</p> <p>‘If we had known sooner then a lot of worry about what was wrong with our child would have been eliminated. Treatment could have started sooner and may have had more impact on our child’s progress. She didn’t start until she was over one year old even though she has type 1. Our child, if treated earlier, may not now need a peg to be fed or a ventilator at night. She may have head control like she once had if diagnosis and then treatment could have started earlier.’</p> <p>‘I think it would allow for planned, more focused treatment rather than emergency trying to investigate symptoms. It also gives families a chance to come to terms and learn about the condition rather than having to do this whilst look after an already sick child. It would also give the various agencies involved more time to create a treatment plan. My daughter was not diagnosed until a few weeks before she died. By the time we had our first meeting about treatment options the best one available was to move to a hospice with her and keep her comfortable. I think with this condition the amount of time you have before symptoms are obvious are crucial.’</p>
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		<p style="text-align: right;">Parent of child with SMA Type 1 receiving treatment</p> <p>'I agree it is a useful tool. However, I feel it should be a choice rather than part of the national screening per se. I didn't know my daughter had SMA or that we were carriers. If we'd had more children knowing the risks I wouldn't have screened.'</p> <p style="text-align: right;">Bereaved Parent of child with SMA Type 1</p> <p>'Screening may cause extra anxieties in the early days of parenthood if parents don't want to know a diagnosis that soon.'</p> <p style="text-align: right;">Bereaved Parent of child with SMA Type 1</p> <p>As someone with SMA (Type II), I strongly believe in newborn screening procedures when it comes to genetic diseases as I think that the information that it provides is far too important to be ignored. Any available chance not taken to improve someone's current/future quality of life, whether via treatment (after considering the risks of said treatment) or just by planning for the future, is a wasted opportunity and, in my own personal opinion, immoral.</p> <p style="text-align: right;">Adult with SMA Type 2</p> <p>In summary: Evidence to date indicates a high level of support from the SMA community and the public for newborn screening for 5q SMA</p>
	<p>Our conclusion</p>	<p>We conclude that a further urgent review of a newborn screening programme for 5q SMA in the UK is imperative due to the development of a life-saving intervention for children with SMA, together with clear evidence from trials that early treatment has a major influence on subsequent functional outcomes, the rapid changes we are seeing with research and the development of other new treatments and the introduction of screening programmes in the USA and other countries.</p>

		<p>We urge the NSC to start this review as soon as the outcome of the NICE appraisal and the potential of a Managed Access Agreement and the outcome of further deliberation about extending treatment in Scotland is known.</p> <p>We request that if other criteria are met, any current population uncertainties should not hold up the possibility of progressing a 5q SMA newborn screening programme. We note, that when used in newborn screening programmes in the USA, the information about <i>SMN2</i> copy number and other tests are being considered as sufficient to predict the severity of an infant's 5q SMA and to establish which children should be treated and which monitored.</p> <p>We ask that if testing is introduced in the UK it would need to be made clear to parents that the test is for 5q SMA, the most common form of SMA with clear information that this test does not cover all possible other very rare forms of SMA.</p> <p>We note that there is a growing body of clinical trial and real-world evidence of the effectiveness of nusinersen treatment for 5q SMA and that the earlier the treatment the greater the impact, with strong indications that the greatest impact occurs if it is started pre-symptomatically.</p> <p>We note that other treatments on the horizon are also indicating that the earliest possible treatment has the greatest impact.</p> <p>We note that there are significant ethical implications and dilemmas posed by newborn screening for 5q SMA. This is especially heightened with a treatment such as nusinersen, which is invasive in delivery (lumbar puncture) and requires further life-long treatment once every four months.</p>
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		<p>We consider it vital that any screening programme is accompanied by a robust and ongoing programme of supportive counselling and accurate information about: what information may be revealed via the screening test; what impact the various types of 5q SMA have on life; what management and treatments are available.</p> <p>We note that evidence to date indicates a high level of support from the SMA community and the public for newborn screening for 5q SMA.</p> <p>Again, we note that at this stage it is not known which children will be able to access nusinersen treatment in the UK. This decision and the possible criteria for a proposed screening programme are critical to any further debate and decision making which, as stated above, we consider to be imperative at this time.</p>
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Please return to the Evidence Team at screening.evidence@nhs.net by 9th September 2018