Public consultation summary document

Organisation /Individual	Feedback	Changes made	Oversight Committee review
[redacted]	The report appears very comprehensive. One area that could be bolstered is the role of clinical genetics in SMA and other genomic newborn screening. If this is not adequately included then the genetic support for genomic newborn screening will be diminished at the expense of the direct and indirect family. We are somewhat biased at [redacted] given our model mostly means that a genetic counsellor usually joins the paediatric neurologist at the first appointment. I think it would be useful to have a genetic counsellor as a member of the working group to contribute to this component of the pathway. I don't think the genetics aspects of newborn screening are being adequately considered. Many of the psychosocial supporting clinicians provided by the MDT do not feel confident addressing the genetic questions that families inevitably have (reproductive carrier testing, pattern of inheritance, implications to other siblings and the wider family, complexities around and facilitating carrier testing and implications to future offspring and reproductive testing). A genetic counsellor can provide support and provide detailed knowledge around the genetic aspects of an SMA diagnosis. These questions are usually raised at the same time of the diagnosis disclosure and access to a genetic counsellor (F2F or via telehealth) is an essential component of care to support the family through a very stressful time. I can see that referral recommendations to clinical genetic services are present but they appear optional or something that can be addressed later issue which is not our experience.	The GDG has already been formed and therefore this feedback cannot retrospectively be actioned. The role of clinical genetics services and genetic counsellors have been expanded in the background of section 9 and with an additional implementation point 9.5.1. whilst also allowing for the fact that not all health jurisdictions have ready access to clinical services. The revised recommendations now state in Section 9: Background *Care, support and targeted information is imperative within the post diagnostic stage for families. Genetic counsellors fulfil a vital role in providing support and addressing the genetic questions that families inevitably have as pertains to a diagnosis of SMA (i.e. on reproductive carrier testing, pattern of inheritance, implications to other siblings and the wider family, complexities around and facilitating carrier testing and implications to future offspring and reproductive testing (ref). Whilst many jurisdictions have conjoined clinical genetics and neurology services to facilitate genetic support at the time of diagnosis, for families living in jurisdictions without these shared services, early referral to clinical genetics centres for review is deemed important (ref again).* Implementation point 9.5.1 (added to finalised draft). Whilst it is ideal that families have support and genetic information from clinical geneticists and genetic counsellors at the time of diagnosis (as part of the multidisciplinary care team), healthcare jurisdictions have variations in access to clinical genetic services. Therefore, clinical referral should occur within appropriate and pre-established local pathways.	Agree Agree
[redacted]	Congratulations on this, it's a great draft. I do however have two concerns: • **Genetic Counsellors (GCs) should be mentioned as specific health care practitioners in the guideline** ○ Families value the education and psychosocial support routinely provided by after a NBS screening diagnosis per our pathway here [redacted] is a Genetic Counsellor who did a study comparing our SMA NBS cohort with the Metabolic NBS cohort from [redacted]	The role of genetic counsellors has been further highlighted through the Guideline in view of the feedback in the following sections. Background section 5: Dependant on health expertise and confidence in disclosing sensitive results to families, other programs have leveraged the experience of trained genetic counsellors or nurses, particularly in regional and remote areas. Recommendation 5.3 clarification We suggest that it is acceptable for a designated healthcare practitioner with support from a paediatric neurologist to disclose a screen positive result to a family.	Agree

	who do not receive genetic counselling. The results demonstrated the benefit of genetic counselling after a NBS diagnosis. [redacted]. ○ There are numerous GCs in Regional Settings available to support local Medical Practitioners when disclosing the diagnostic results. This has not been mentioned in this document and I feel that it needs to be. A regional GC is the ideal person to provide follow up support and education for the family, as well organising cascade testing and advice for future pregnancies. This would be done by a GC rather than a Clinical Geneticist, as is the case in our pathway. Aside from these points, I think it is a very exciting to see this come together. Congratulations again on a wonderful document!	Grade of recommendation Conditional, Grade 2C *The designated healthcare practitioner will vary between health jurisdictions and may include general practitioners, paediatricians, specialist nurses and/or **genetic counsellors**. These individuals should have training and expertise in disclosing screen positive SMA results to families.* In Section 8, Rationale and Impact There was a unanimous consensus that the preference was for result disclosure to be the responsibility of specialist medical practitioners (usually paediatric neurologists) with expertise and knowledge in the condition and next steps to expedite treatment, aligning with the evidence of parents valuing early specialist input. However, the GDG agreed that processes for result disclosures were jurisdictionally dependent, and that medical practitioners such as **genetic counsellors** nurse specialists and non-specialist medical practitioners could also be well placed to disclose and counsel on the results. For these professionals, the evidence showed that access to and advice from specialist services, enabled a streamlined and effective disclosure process. Recommendation 7.2 We suggest that the number of healthcare practitioners at the first clinic visit for diagnostic evaluation (following screen positive disclosure) should be limited to those necessary for information disclosure and may include the information provider (usually a paediatric neurologist or paediatrician), and ideally support from representatives of the clinical genetics service (**geneticists and/or genetic counsellors**) and/or medical social work and/or psychological services. Recommendation 7.7 We recommend that all families receiving a diagnosis of SMA for their newborn, through a newborn screening program should be provided with the contact details of a designated healthcare practitioner who can direct a response to their queries. Grade of recommendation Strong, Grade 1C *The designated healthcare practitioner will vary between health jurisdictions and may include but are not limited to paediatric neurologists, paediatricians, genetic counsellors or specialist nurses.* Recommendation 8.3. We suggest that if circumstances dictate and dependent on individual (family and child related) factors, it is acceptable for a designated healthcare practitioner (such as a paediatrician, general practitioner, specialist nurse, genetic counsellor) with support from a paediatric neurologist to disclose a diagnostic result to a family. Grade of recommendation Conditional, Grade 2C	
	At least one GC should be present on the Guideline Development Group Given the importance of the role, the Guideline Development group would benefit from a GC's detailed subject-specific knowledge in the area.	The GDG has already been formed and therefore this feedback cannot retrospectively be actioned.	

			Agree
[redacted]	The feedback from one colleague was to please replace “New Zealand” with “Aotearoa New Zealand” in all documents P25 of the Guideline “… internationally developed SoC for SMA..” - References 25 and 26 are quoted. I wondered whether specifically for SMA the reference 50 and PMID: 29305137 (which is not listed as a reference at all) would be more appropriate. P59 of the Guideline - there are two recommendations 10.15 and two recommendations 10.17 – 1 of each should be 10.14 and 10.16, respectively. Page 106 Fig 4 – SMN2 produces 6 hexagons worth of full length SMN protein in a healthy individual but only 3 in a SMA patient – not sure what this is meant to indicate? Again, my respect and congratulations for your amazing work!	Aoteroa has been added All references are now aligned The Figure has been redesigned to be representative	Agree Agree Agree
[redacted]	[redacted] in the Newborn Bloodspot Screening (NBS) decision-making pathway, which ensures national consistency in partnership with states and territories 2. Spinal Muscular Atrophy (SMA) is a condition listed for screening as part of the NBS program 3. Children born [redacted] with SMA would be cared for in partnership with sub-specialists based at institutions such as [redacted]	No changes required	Agree
[redacted]	Health and Social Policy Branch has reviewed the draft Guideline and do not have any specific feedback. [redacted] is committed to participation in the national process underway to achieve national consistency for NBS, and I commend you and your team on your work to support these principles. I look forward to reading the final version of the guideline when published.	No changes required	Agree
[redacted]	Upon review of both the National Recommendations for Newborn Screening in Spinal Muscular Atrophy in Australia and New Zealand Guideline Document, as well as the National Recommendations for Newborn Screening in Spinal Muscular Atrophy in Australia and New Zealand Administrative and Technical Report, there were noted areas of repetition that may be truncated or condensed to enhance accessibility and readability. Specifically, but not exhaustively: 1.Grading the direction and strength of evidence-based recommendations Page 85 Page 84 While not a word-forword repetition, suggest limiting to one document 2.Stakeholder consultation activities – systematic observation form evidence on page 89 Systematic observation forms to collect expert evidence on page 77 Text repeated wordfor-word	As per NHMRC guidance, grading process is preferable in both documents	

	3.Healthcare practitioner survey (modified Delphi process) Page 91 Page 79 Text repeated wordfor-word 4.Western Australia has already been screening for SMA for over a year. It would benefit the Guideline to reference them and/or adopt some of their		
[redacted]	Acknowledged receipt of invitation letter but did not provide formal feedback.	No changes required	
[redacted]	[redacted] supports the implementation of the National Recommendations for Newborn Screening in Spinal Muscular Atrophy guideline.	No changes required	Agree
[redacted]	• P21 whilst I agree that 'back up gene' is not an ideal term for SMN2, to me the phrase 'nearby related gene' is a bit confusing, so I wonder if it would be clearer to say 'related gene… located near SMN1'?	This has been corrected and now reads related gene, located near SMN1.	Agree to all feedback
	• P25 Population – I know it is mentioned further on, but I wonder whether it would be good to mention early in the document that SMA affects all populations/ethnic groups (albeit at varying frequencies)	This has been incorporated and now reads Guideline purpose, scope, population and settings: Whilst incidence and prevalence varies between groups, SMA affects all ethnic populations.	

	• I note that you have varyingly referred to absence/loss of SMN1 as 'deletion' throughout the document
	  • I suggest that you are consistent
	  • In most places throughout the document I think it is most correct to avoid the term deletion – as this implies mechanism for the loss of SMN1, whereas the testing that we do is just quantitative and only tells us whether SMN1 is present, not how it was lost. I understand that a significant proportion of patients are thought to have lost their SMN1 through gene conversion rather than deletion per se
	  • Suggest using loss, absence, deficiency.
		Whilst the screening assays are targeted at biallelic deletion of exon 7 in SMN1 and have thus remained the same, where appropriate, absence of exon 7 on SMN1 has been added.	

	• Suggest adding 'clinical' to geneticist throughout the document (where that Is what you mean!) – including the diagram	The word clinical has been incorporated throughout the document.	
	• P39 I think it would be useful to add that sometimes testing of parents is suggested to try to work out why there is a false positive or uninterpretable result	This has now been added as a good practice point which reads Good practice point 4.3.1. Venous samples from parents for *SMN1* quantifications purposes may be required to inform the aetiology of a false positive or uncertain result for the newborn.	
	• P42 – I think the term 'responsible medical practitioner' is ambiguous – I presume you mean responsible for the patient rather than someone not irresponsible!	This has now been changed to designated healthcare practitioner throughout the document. This has been changed particularly in recommendation 6.1 Venous sampling for quantification of *SMN1* on whole blood.	

	• P46 – there are a few places where you say 'venous sampling for SMN1' – I don't think this makes sense? Should be it venous sampling for quantification of SMN1? – and then similarly, venous sampling for determination of SMN2 copy number? • P100 – in 1st paragraph – you mention the scenario of two sequence variants – but they need not necessarily be homozygous – is more correct to say 'biallelic sequence variants' (could be homozygous or compound heterozygous). • P114 I think the more correct term is 'reproductive genetic carrier screening' (but noting that the MBS uses 'testing' not screening) • P117 last paragraph & p119 – I don't think the sequence variant needs to be in exon 7 – there are recurrent variants in exons 1,3 & 6 in particular • P161 you mention the phrase 'done incorrectly' - I am not sure that this is a binary thing - right or wrong – I suspect it is better to reword this in a way that says we want to deliver this devastating news in the most constructive/least traumatic way possible rather than correct vs incorrect • I note the use of the term 'allied therapist' in several places throughout the document – I am more familiar with - 'allied health therapist/specialist/professional'? • I wasn't sure whether I was looking in the right place for ref 24, 25 and 26 below – which don't appear to match the numbered ones at the end of the document	Venous sampling for determination of *SMN2* copy number on whole blood OR repeat dried blood spot for confirmation of *SMN2* copy number. This has now been altered. This has now been changed to reproductive carrier testing. Please see ICER comments This has been changed and now reads in Rationale and Impact Section 5:The evidence reported that some families felt that the information given at this juncture set the tone of the healthcare journey and could challenge family perception, engagement and trust in care thereafter The term has now been rewritten as allied health therapist throughout the document. The references have been realigned	
[redacted]	Section 5: Disclosing a screen positive result to families [redacted] recommends that written information, either as a standalone document or by referral to a website, is provided to parents immediately following the disclosure phone call. This information should be available in an accessible format and in different languages. The 2021 Census shows that a language other than English is used in 28% of households [redacted] (Cultural diversity: Census, 2021 | Australian Bureau of Statistics (abs.gov.au)). We suggest the written information provided to families includes plain language information for recommendation 5.10 advising families to contact the medical practitioner if the following are noted in the newborn/infant: change in movement, feeding, or breathing pattern, change in voice or weak cry, increased	Additions have been made to reflect the feedback The GDG highlighted the need to standardise information provision (through verbal and written means) and highlight signs and symptoms of clinical deterioration, to mitigate clinical risks to the child. Implementation point 5.101.1 We suggest that written information is provided to families either as a stand-alone document or by referral to a website so that families can access reliable and well curated information at the point of screen positive disclosure, inclusive of red flag signs and symptoms that necessitate immediate clinical review. This information should be in an accessible format and made available in a range of languages.	Agree

	fatigue without increased activity, decline or loss of function in previously attained motor ability or failure to show progress in expected motor ability, abdominal breathing and/or failure to thrive. It is unlikely that parents will be able to remember or assess clinical signs without written resources and accessible support from a health professional. Alternatively, this recommendation may need to be simplified to alerting a health professional if parents have any concerns about their newborn rather than listing the clinical signs which may be too burdensome for newborn parents who have received a positive screening result.		Whilst 42 days is within the evidence base, there is also evidence that each day without treatment counts. The completion of the screening-diagnosis to treatment cycle was felt to be feasible within 30 days and the wording adjusted to reflect this.
	Section 3: Confirming the diagnosis of spinal muscular atrophy. We recommend the timeline for diagnostic results is clearly stated in the guidelines. For example, results are required such that treatment can begin by 6 weeks of life, if this is consistent with the evidence provided below in Section 3. The timelines appropriate for completion of all diagnostic tests for SMA (including SMN1 and SMN2 copy number) should be as short as possible, without compromising the accuracy of the process. This is emphasised by the fact that children diagnosed and started on SMN augmenting treatment by 6 weeks of life have a higher probability of following normal motor development trajectories, independent of SMN2 copy number. Therefore, time to diagnosis and subsequent treatment appears to be a substantial modifier of health outcomes for these children.	Recommendation 3.8 Consensus based recommendation We suggest that diagnostic test results (including *SMN1* and *SMN2* copy number) should be available to clinical services ideally within 30 days of birth (and mandated before 42 days of birth) to enable timely treatment. Grade of recommendation Conditional, Grade 2B	
	Section 4: Managing uncertain, false positive and false negative screening results We suggest that lessons or insights derived from the 'root cause analyses' of false positive/false negative or uncertain results are shared between Australasian Newborn Bloodspot services so that common issues and errors can be identified. This would be in addition to the knowledge exchange activities described below in Section 4. The Guideline Development Group (GDG) highlighted the need to undertake knowledge exchange activities across Australasia of the limitations of newborn screening for SMA, to emphasise the necessity for prompt referral to clinical services for symptomatic children due to the potential for false negative cases (due to the inherent limitations of the target assay, human/system error or probe binding issues).	Implementation point 4.7.1. We suggest that lessons or insights derived from the case review of false positive/false negative or uncertain results should be shared across Australasian Newborn Bloodspot services so that common issues and errors can be identified.	Agree
	Section 7: Information provision to families during the diagnostic evaluation of a screen positive newborn and after confirming the diagnosis of SMA We recommend nationally consistent and up to date information is available to all families who receive a screen positive newborn result and a diagnostic positive result based on the evidence below from Section 7. The evidence showed that families struggled to find sources of information other than their doctor and the GDG acknowledged that clinics could leverage local and national support groups to augment information provision.	Section 7 background: Families often describe a period of information seeking between screen positive disclosure and diagnosis, associated with feelings of distress and confusion. Well curated and reliable sources of information at screen positive disclosure are considered vital to bridge the information gap and provide accurate counsel. Recommendation 7.6	Whilst the committee agreed that educational resources were important (recommendatio

		We recommend that families receiving a diagnosis of SMA for their newborn, through a newborn screening program, should be directed to high quality and reliable educational resources that support information provision on the implications of the diagnosis and potential treatments for their newborn. Grade of recommendation Strong, Grade 1C *Educational resources provided should reflect the contemporary treatment and care landscape and be nationally consistent.*	n 7.6 reworded) they felt that suggestions to update to existing support group websites were outside the scope of recommendations and would be a barrier to national implementation, however, this point was acknowledged in the affect on practice, section 7 (see below)
	The GDG highlighted through clinical experience and consensus that a tailored program of information provision was required, paced and adjusted according to the preferences and circumstances of the family. We recommend there is a smooth process to transition the newborn from screening, diagnosis and post diagnosis across clinical care, with information and resources and psychosocial support throughout. The process should 3 Guideline Feedback recognise each family will be at different stages of understanding the information and be tailored to each families' unique needs based on the information below from Section 7. The evidence showed that there are gaps in current practice in communication, information and support available to families. Benefits of high quality, accurate and tailored information provision were considered by the GDG to encompass many levels including improving therapeutic decision making for families and clinicians, improving access to appropriate support, increasing family wellbeing and satisfaction with care and empowering families to be active participants and engage in the healthcare process for their child.	Guideline unchanged	The oversight committee agreed that this point had been considered throughout the recommendations in section 7 and did not require changing.
	Section 8: Delivering the diagnosis and supporting families as they receive the diagnosis of SMA Consistent with Section 7 and recognising the intent of the GDG in addressing the psychological and support needs of families, we recommend all families either have a psychosocial support healthcare professional present at the appointment or receive a phone call offering psychosocial support to the family after the results disclosure.	Guideline unchanged	Whilst this is the gold standard, the Oversight Committee agreed that health jurisdictions varied in access to psychological services. Psychosocial support was suggested throughout recommendations, to

			acknowledge its importance in the model of care but was not mandated to also balance feasibility across Australasia and in reflection of the evidence base grading.
	Section 10: Treatment planning and initiation for newborns and infants diagnosed with SMA through newborn screening programs We suggest that written information or website information is provided with Recommendation 10.9 where medical practitioners will explain to families and document the potential benefits, risks, uncertainties of SMN augmenting treatments and need for long term surveillance. This information must be available in accessible format and in different languages. The recommendations 10.15 onwards refer to the newborn diagnosed with SMA "through newborn screening" where this terminology has not been used in the other recommendations. It is unclear whether the clinical recommendations apply to newborns diagnosed with SMA regardless of whether it is through newborn screening or clinically following a negative newborn screen. Guideline impact	Guideline changed with addition of good practice point 10.9.1 We suggest that written information as a stand alone document or direction to a well-curated, reliable and up to date website is provided to families that will inform them on the potential benefits, risks, uncertainties of SMN augmenting treatments and the need for long term surveillance. The information should be in an accessible format and ideally provided in different languages.	
	For [redacted], and likely other jurisdictions, the guideline will alter the diagnostic pathway, shifting it from a clinical diagnosis triggered by clinical signs to a newborn screening triggered diagnosis. The implementation of additional newborn and reproductive screening will increase the demand for both reproductive counselling and pre-implant genetic testing.	This is now acknowledged in Section 7: How recommendations may affect practice section which now reads; The recommendations complement current practice, that encourages family centred care for families within a multidisciplinary team setting, so the GDG agreed that for some jurisdictions there would be no substantial resource impact. However, with the alteration of the diagnostic pathway through newborn screening, the demand for reproductive counselling and preimplantation genetic counselling would increase in certain health jurisdictions. The GDG acknowledged that members of the wider multidisciplinary team (extending to patient organisations) could augment roles as information and support providers dependent on jurisdictional resources and capacity, and that their websites could be leveraged to provide targeted and reliable information for families receiving a diagnostic result.	
	Barriers and facilitators of implementation recommendations Barrier to implementation: Lack of appropriate resources for patients/families. For example, the Australian SMA advocacy and support group website will need resources specific for families when a positive screening result and diagnostic result is received. Spinal Muscular Atrophy: Causes, Symptoms, & Treatment (smaaustralia.org.au). Facilitator of implementation: Jurisdictional consistency in implementation is preferable, and identification of a mechanism for key stakeholders in each jurisdiction to coordinate and provide consistent communications will support successful implementation of the recommendations across screening, diagnostic and post diagnosis care.	Recommendation 7.6 We recommend that families receiving a diagnosis of SMA for their newborn, through a newborn screening program, should be directed to high quality and reliable educational resources that support information provision on the implications of the diagnosis and potential treatments for their newborn. Grade of recommendation Strong, Grade 1C *Educational resources provided should reflect the contemporary treatment and care landscape and be nationally consistent.* We suggest that written information as a standalone document or direction to a well-curated, reliable and up to date website is provided to families that will inform them on the potential benefits, risks, uncertainties of	Whilst the committee agreed that educational resources were important (recommendation 7.6 reworded) they felt that suggestions to update to

		SMN augmenting treatments and the need for long term surveillance. The information should be in an accessible format and ideally provided in different languages.	existing specific support group websites were outside the scope of recommendations and would be a barrier to national implementation. Whilst the oversight committee acknowledge this point, for the purposes of brevity and signposting, NBS for SMA was considered the most appropriate title, with the contents ascertaining that it was taken as a healthcare journey for the child
	Overall feedback The title of the guideline does not reflect the breadth of the content. Suggest the title includes reference to 'diagnosis' and 'post diagnosis' in addition to screening to ensure it captures the attention of the appropriate stakeholders beyond the newborn bloodspot screening laboratories. This will align with the Executive Summary, 'to span the entire healthcare journey of the newborn'.	Title unchanged	
	Technical report No feedback Family fact sheet No feedback Additional feedback		
	The draft guidelines recommend five yearly review and update. We suggest adding an option to review the guideline should new practice changing evidence become available.	Reworded and now states The Guideline should be reviewed in 5 years of publications or sooner if the screening, diagnostic or clinical landscape changes in the interim, updated to reflect and respond to new evidence from research, clinical practice and changes in community needs, values and preferences.	Agree

[redacted]	Guideline document Really good. Obviously very thoroughly researched, proof read and edited. Few notes. On page 20, states: "Decentralisation of newborn screening in Australia and New Zealand may give rise to regional differences in newborn screening programs"New Zealand has a centralised NBS programme. Not sure if they are trying to say it that it's not centralised across the two countries? May be better to say: "Decentralisation of newborn screening in Australia and a separate system in New Zealand may give rise to regional differences in newborn screening programs" On page 21: "In 2022 and 2023, the federal governments of Australia and New Zealand respectively…"Federal government is not a term used in NZ. Would suggest deleting the word federal to just say: "In 2022 and 2023, the governments of Australia and New Zealand respectively" On page 25: "It is made to be flexible and adapted to conform with available resources and capacity on a state/territory level across Australia and New Zealand."Would suggest: "It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and New Zealand. On Page 54: "Recommendation 9.9.Consensus based recommendationWe suggest that newborns undergo neurophysiological assessments within a reasonable time of diagnosis, including collation of compound muscle action potential (CMAP) +/- electromyography (EMG), to obtain predictive information on disease course. Strength of recommendation Conditional, Grade 2C"[redacted]. Technical report • Previous comments about the executive summary in the other document regarding inclusive language [redacted] is all true in this one too.	This has now been amended and reads Decentralisation of newborn screening in Australia and a separate system in New Zealand may give rise to regional differences in newborn screening programs This has been deleted and reworded as per suggestion This has been deleted and reworded as per suggestion The variability of access to equipment and personnel to complete these assessments has been acknowledged in the Guideline and no change required	Agree Agree Agree

	• Under Risk Assessment pg 103: A further risk not mentioned that could be consider that's no specifically mentioned is that the introduction of genetic testing to the NBS programme may lead to disengagement with the overall NBS programme, particularly for indigenous populations who may have additional concerns around data sovereignty of genetic information and implications [redacted]. This could be considered in the context of point 3 "the risk of widening health inequalities across Australia". Also both point 2 and 3 should be "… across Australasia". • Under Dissemination and Implementation plan pg 105: No mention of implementation in NZ. Add a sentence "In New Zealand this is overseen by the national Newborn Metabolic Screen Programme". Otherwise all good.	-Sentence added	

[redacted]	General feedback Clinical services have already absorbed 3 SMA treatments, increased patient numbers due to survival, increased complexity in treated symptomatic patients, coordination of care, coordination of treatment programs and support and managing the care. NBS programs have also increased demand for clinical services, critically urgent review and initiation of treatment & the intense monitoring post treatment. There has been no additional resourcing of services to support the increased clinical workloads. It remains challenging to provide SOC to patients with NM disorders. To implement the SOC for NBS screening programs, clinical services need additional funding to build capacity, workforce, succession planning. Screening feedback Health literacy ... non English speaking backgrounds, cultural considerations, Temporary Visa status - NBS offered to all infants, regardless of Medicare status and eligibility, however access to care and PBS funded treatments is restricted. Families may not be able to afford access to care or genetic testing, genetic counselling etc. How is this managed in other NBS programs? Diagnostic feedback Variability between in states for Tier 2 testing SMN1 & SMN2 confirmatory testing timeframes. [redacted] for 2nd tier .. - 7-10 days turn around. Much quicker for other states - [redacted]	-Resourcing has been addressed in the implementation protocol. The Guideline is intended to inform and guide but does not replace clinical reasoning or acumen. It is linked with and thus do not replace the National Screening Policy Framework (34) and internationally developed Standards of Care for SMA.(35, 36) It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and Aotearoa New Zealand. As such, it has been developed within the current health policy framework of these two countries and the parameters of the Guideline do not specifically address reimbursement pathways for children with SMA (diagnosed through newborn screening) who are not eligible for subsidised or publicly funded health services or treatments.	

	[redacted] - logistics with timely access to care and confirmatory testing - will likely cause delays - maybe outside of the recommended timeframe of 7-10 days. Clinical feedback Our local experience has shown that whilst NBS is done on most patients, however not all have Medicare. 50% of NBS this year. Immigration /Visa status impacts access to clinical care and treatment options. Hospital systems, service demand/capacity restraints. Impact on clinical services .. demand, survival, critical timeframes , clinical services struggle to juggle and absorb workload to provide diagnostic, treatment and ongoing clinical care. Clinical services need additional resourcing / staff to deliver services. SMA care has changed dramatically in the last decade, however clinical resourcing & funding of service has not responded to this demand. Guideline potential implications Improved awareness and understanding. Consumer expectations ... logistical and systematic barriers which impact the delivery of clinical services. Recognition for the importance of SMA care, timely access to treatment. Hopefully - appropriate resourcing of services, additional funding, capacity building, succession planning Barriers and facilitators Inequity in care still exist - Treatment eligibility - no Medicare - can't access PBS funded treatments, can't access NDIS supports to meet SOC recommendations. Insurance status - variability ... SMA treatments are high cost, they won't necessarily be covered by insurance. Family who have NO private health insurance and no Medicare. Challenges - NBS positive, confirmatory genetic testing, unable to access treatments; family with no insurance to cover treatment or care. Will State based health systems absorb the cost, how do we advocate for compassionate access to treatments ?	While the SAC recognises the geographical differences between states, this Guideline has been developed as a best practice protocol for NBS for SMA. Addressed in Scope: The Guideline is intended to inform and guide but does not replace clinical reasoning or acumen. It is linked with and thus do not replace the National Screening Policy Framework (34) and internationally developed Standards of Care for SMA.(35, 36) It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and Aotearoa New Zealand. As such, it has been developed within the current health policy framework of these two countries and the parameters of the Guideline do not specifically address reimbursement pathways for children with SMA (diagnosed through newborn screening) who are not eligible for subsidised or publicly funded health services or treatments. The Guideline is intended to inform and guide but does not replace clinical reasoning or acumen. It is linked with and thus do not replace the National Screening Policy Framework (34) and internationally developed Standards of Care for SMA.(35, 36) It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and Aotearoa New Zealand. As such, it has been developed within the current health policy framework of these two countries and the parameters of the Guideline do not specifically address reimbursement pathways for children with SMA (diagnosed through newborn screening) who are not eligible for subsidised or publicly funded health services or treatments. Implementation point 10.1.1 Australia and Aotearoa New Zealand treatments for SMA are subsidised by the publicly funded healthcare system for children who meet eligibility criteria. Reimbursement structures and options for treatment vary across the two countries. For children who are not eligible to access subsidised treatments on the basis of residency status or other factors, treatment and care pathways require interrogation on a case-by-case basis. ‘	

[redacted]	General feedback Slide - What is NBS for SMA Blue circle Please correct 2 spelling errors "manging" to managing and "screeing" to screening	-Changes made according to feedback	

[redacted]	General feedback Too long block information Information repeated Difficult to follow and interpret Needs some table approaches/flow charts Yes/no direction of treatments Target audience may not be experts in this field Clinical feedback Lack of clinical pathways from diagnosis for example :- Maternity hospitals (birth hospitals) will receive the positive result in [redacted] and this Information will be to the to local paediatrician No mention of specialist nursing teams Clinical Nurse consultants Nurse specialists	Inactionable comments This has been acknowledged in implementation point 2.7.1 Good Practice point 2.7.1. We suggest that the newborn screening for SMA program will establish a clinical referral pathway for newborns who screen positive for SMA. A positive newborn screening result may also be verbally relayed to a relevant listed healthcare practitioner (that is or has been involved in the care and management of the child such as a general practitioner or paediatrician). Recommendation 5.3 Consensus based recommendation	

	Where does the role of the Primary Health Care provider fit in (GP) <u>Potential Guideline impact</u> Resourcing On going funding Access to services in the "recommended" time frames Considerations of funding for non-eligible families	We suggest that it is acceptable for a designated healthcare practitioner with support from a paediatric neurologist to disclose a screen positive result to a family. Grade of recommendation Conditional, Grade 2C *The designated healthcare practitioner will vary between health jurisdictions and may include general practitioners, paediatricians, specialist nurses and/or genetic counsellors. These individuals should have training and expertise in disclosing screen positive SMA results to families.* *Support as defined in this recommendation can range from exchange of advice, information (verbal and/or written) or a formal offer to be part of the screen positive disclosure, alongside the designated healthcare practitioner.* Implementation point 10.1.1 Australia and Aotearoa New Zealand treatments for SMA are subsidised by the publicly funded healthcare system for children who meet eligibility criteria. Reimbursement structures and options for treatment vary across the two countries. For children who are not eligible to access subsidised treatments on the basis of residency status or other factors, treatment and care pathways require interrogation on a case-by-case basis. The Guideline is intended to inform and guide but does not replace clinical reasoning or acumen. It is linked with and thus do not replace the National Screening Policy Framework (34) and internationally developed Standards of Care for SMA.(35, 36) It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and Aotearoa New Zealand. As such, it has been developed within the current health policy framework of these two countries and the parameters of the Guideline do not specifically address reimbursement pathways for children with SMA (diagnosed through newborn screening) who are not eligible for subsidised or publicly funded health services or treatments.	

[redacted]	<u>General feedback</u> fantastic, well thought out <u>Clinical feedback</u> Recommendation 9.5 (referral to genetic counselling) does not seem to incorporate an understanding that some areas of mainstreaming genetic counselling is growing and it may not necessarily be a 'clinical genetics unit' that	This is a point that has been considered by the SAC and recommendation 9.5 has been changed and now reads We suggest that families of newborns diagnosed with SMA through newborn screening programs should be offered referral to, and review for genetic counselling and cascade testing (which may include referral to a clinical genetics service),	

	provides this counselling. There may be genetic counsellors within the neuromuscular multi-D team who will provide this. Would it be easier to say refer for genetic counselling and cascade testing (which may include referral to a clinical genetics unit)???		
[redacted]	General feedback • On review, the guideline appears comprehensive and aligns with the work by policy makers in states and territories and the Commonwealth. • Keen to understand how these guidelines when finalised will be disseminated, promoted and used to support SMA integration into newborn bloodspot screening (NBS) – noting it is already part of NBS programs across the country. Assume this will be via s/t and hospital networks to reach clinicians, consumers etc?	An implementation document has been provided as a separate file and is located on the website, with a link provided in the Guideline document under the section of future directions; dissemination and implementation of recommendations within the Guideline.	
[redacted]	General feedback We have sought expert clinical feedback on the guideline. The advice is, while the recommendations are reasonable, they are mostly not of direct relevance to GPs. As such, it would not be appropriate for [redacted] to be listed as an endorsing organisation. We therefore respectfully request you remove [redacted] from the list of endorsing organisations.	-no change needed [redacted]	
[redacted]	General feedback The consensus-based recommendation grading system detailed on pg 90 (i.e., 1A-2C) would be useful to include in the 'list of recommendations' on pg 28 to help understand the grading for these recommendations, and minimise confusion with the evidence-based recommendation grading system. Screening feedback A few recommendations are a little redundant and/or may overlap with other guidance already available/applicable to all NBS conditions, e.g., Recommendation 1.1 is national policy in Australia that has already occurred through an alternative recommendation pathway and has already been	Although this is a fair point, on the weight of feedback, the SAC felt that the location of the Table 5 with the grading system was appropriate. However, a hyperlink has been added between the pages for reference (inserted between List of recommendations and Step 8). Whilst recommendation 1.1. is true, the SAC felt that it was still important to keep within the Guideline as other jurisdictions (outside of Australasia) continue to assess saliva and whole blood to implement NBS for SMA. The SAC felt that Recommendation 1.8. was still within the scope of NBS for SMA.	

	implemented, and Recommendation 1.8 – does this duplicate existing guidance on taking bloodspots prior to transfusions? Also, if this recommendation is targeted at sample collection staff it differs from almost all of the other recommendations and it is not clear that this is a key audience for the guidelines. The use of the term "screen positive" is used differently in different parts of the guidelines and wording may need to be clarified – Recommendation 1.7 refers to the "screen positive" result being communicated as just the SMN1 result, which does not align with the definition in Recommendation 2.3 being both the SMN1 and SMN2 results defining a "screen positive".	This has now been clarified and section 2.3 now reads We recommend that when *SMN2* copy number is known to be > 4 at the time of initial newborn screen identification i.e. in the absence of exon 7 on *SMN1*, this is **not** designated as a screen positive result. Section 1.7. We recommend that a screen positive result should be communicated to clinical services when the *SMN1* screening result is available (independent of the availability of *SMN2* copy number on screening assays), to reduce timelines to diagnosis and treatment.	

[redacted]	General feedback Thank you for such a comprehensive guideline and for thinking so deeply about the experience of patients and families. The only feedback I would like to give and have considered is the inclusion of referring or at least making families aware of the existence of SMA Australia, and other support organisations like Genetic Support Network of Victoria and Genetic Alliance Australia. We have learnt that unless this is explicit it is often overlooked. Section 9 I believe is where this would be most relevant.	The SAC has discussed this feedback and felt it is not prudent to incorporate specific advocacy group names. We have titled these within an umbrella term of support organisations, with the clinician role to identify the most appropriate in terms of the family's needs and preferences. This has been added into the definition section of the Guideline under the title 'The definition of advocacy services' and states the GDG recognised that a variety of international, national and jurisdictional services exist for children with SMA and their families. For the purpose of the Guideline these have been grouped under the terminology of advocacy services. We leave it to the discretion of relevant healthcare practitioners to direct families to the most appropriate services based on individual needs and preferences.	

[redacted]	Screening feedback • **The definition of newborns, infants and children with SMA (pg 25, 100).** The Reading the Guideline the Population sections of the guideline outline that NBS for SMA could occur after the defined period for newborns (<= 28 days), expanding the NBS testing period out to 12 months of age. We note that the Guideline Development Group (GDG) defined the cohorts of newborns and infants with children. Although this seems to contrast with recommendation 3.8, regarding diagnostic *SMN1* results being delivered within 30 days of birth, we recognize, as outlined in the Guideline, that in some circumstances this timeframe may not be logistically practical. • Recommendation 1.2	No changes required.	

	As outlined in the guidelines, recommendation 1.2 reflects that 95% of newborns with SMA is due to homozygous deletion of exon 7. The other 5% is made up of a compound heterozygote genotype, biallelic pathogenic sequence variants or SMA not due to SMN protein deficiency. This approach is consistent with other countries including Canada (Groulx-Boivin et al., 2024). As outlined in the guidelines, patients affected by SMA not picked up by newborn screening would follow the normal clinical pathway. We anticipate future review of the guidelines would include a consideration of ways to incorporate this 5% group into newborn screening, particularly as testing technologies advance.	We have added a sentence in Future directions: the evolution of genomic capabilities in newborn screening. The added sentence states. "This is particularly important for the 5% of children who would not be identified through current NBS for SMA for practices.	
	• Recommendation 2.4 (pg 33,130) We recognize the complex question regarding timing of result disclosure of an *SMN1* positive screening result in relation to the result of determination of *SMN2* copy number. The reasons outlined in the guidelines for this decoupling reflect that *SMN2* copy number determination is not a confirmatory test; as a prognostic marker is not absolute and can vary depending on the methodology used. Clinical presentation is the absolute measure of disease severity. The approach adopted by the guidelines is balanced regarding the timing of the *SMN1* screening result which still incorporates guidelines on the utility of *SMN2* copy number as a prognostic marker (recommendation 2.1, 2.2, 2.3, 2.4, 2.5, 2.6).	No changes required.	
	<u>Diagnostic feedback</u> **General comment on technique of screening.** As noted in Mercuri et al., (2018), the gold standard of SMA genetic testing is a quantitative analysis of both *SMN1* and *SMN2* using multiplex ligation-dependent probe amplification (MLPA), quantitative polymerase chain reaction (qPCR) or next generation sequencing (NGS). The guideline summarized a study by Tavares et al., (2023) that concluded real-time PCR methodologies are accurate and cost effective. This study used MLPA as the confirmatory second test. In a systematic review of NBS programmes for SMA, Cooper et al., (2024) found that most programmes used RT-PCR or RT-qPCR as the index test method, with most programmes using MLPA as the confirmatory test. We agree with the need for flexibility in the guidelines including of the technique employed – to allow for the possibility of advances in technology associated with testing. As mentioned in the guidelines, the accreditation for tests will be governed by the usual regulations for diagnostic laboratory clinical testing accreditation.	No changes required. No changes required. No changes required.	

	• Recommendation 3.4 (pg 35, 140) We strongly agree with the need of orthogonal validation utilizing a different methodology for diagnostic testing. This will aid in the robustness of the test overall and decrease the chance of false positives. This was evident in the systematic review of newborn screening programmes by Cooper et al., (2024) with in most programmes, the index test method being RT-PCR and the confirmatory test MLPA (refer to Table 1, Cooper et al., 2024). • Recommendation 3.8 We strongly agree with the need for timely screening and diagnostic results, given the implications for clinical care. Newborn screening directly addresses issues relating to delayed diagnosis in the absence of screening (Nishio et al., 2023 review; Lin et al., 2015). The recommended turnaround time of the diagnostic tests should be regularly reviewed with new advances in methodology. Our understanding is that 30 days is feasible in terms of current timelines – approximately 2 weeks for *SMN1* NBS and 8-10 days for *SMN2* copy number determination. • Recommendation 3.9 We agree with this statement, particularly in relation to accurately detailing the method for copy number determination. Additionally, the number of repeats >4 is important for informing phenotype severity (Prior et al, 2020). The information regarding methodology is also important in terms of false positives and negatives. We encourage these conventions to be incorporated into internal diagnostic laboratory policies regarding SMA testing and reporting. <u>Clinical feedback</u> • Recommendation 5.3 / 8.2 / 9.7 / 10.10 / In the guidelines and literature there is a strong emphasis on the need for a multidisciplinary approach to the management of SMA patients. Part of this relates to access to specialised neurology services and clinical genetics services when SMA patients are referred for further genetic testing. We note the access to such services can be challenging in outer regional, remote and very remote parts of Australia which creates issues of equity of access for all Australians including	No changes required. This has been reinforced by the addition of a statement within the future directions section which now reads The Guideline should be reviewed in 5 years of publications or sooner if the screening, diagnostic or clinical landscape changes in the interim, updated to reflect and respond to new evidence from research, clinical practice and changes in community needs, values and preferences. This is particularly pertinent as evolving screening, and diagnostic assays change the time to confirmation of SMA. No change required We thank the reviewer for these insights and have incorporated these barriers to equity in the executive summary as a rationale for the need for a pan-national Guideline.	

	Aboriginal and Torres Strait Islander patients in remote areas. For example, Best et al., (2021) identified barriers of access to clinical genetics and genomics, including current service model designs which centre on urban areas, and limited investment in rural areas. Workforce capacity and capability were also raised including the lack of capacity to engage with genetics specialists. A study by Baazeem et al., (2023) found most tertiary hospitals in Australian cities were in major centres (72% in Sydney for NSW; 82% in Melbourne for VIC; 57% in Brisbane for QLD). We encourage investigation of Telehealth as one possible solution for access to specialist neurology services (as indicated in Recommendation 5.3 and Recommendation 8.2 where travel is not feasible. A recent study (Marne et al., 2023) evaluated a neurology outreach programme to aid in paediatrician training in neurology via video-conferencing and was found to be both accepted and effective. In relation to health access for Aboriginal and Torres Strait Islanders, there are general barriers that contribute to health inequities, including lack of transport, waiting times and a lack of culturally appropriate health information and materials (Australian Institute of Health and Welfare 2024). We note in the recent Health Technology Assessment Policy and Methods Review Recommendation 1: Creating a more equitable system for First Nations peoples and Recommendation 2: Providing equitable access to medicines for paediatric patients. • Recommendation 9.5 [redacted] supports this recommendation and that referral occurs in a timely fashion. This is consistent with current practice, where referral to a specialist genetics service can provide families with expert advice regarding cascade screening testing and recurrence risk. Involvement of genetic counselling at the time of SMA diagnosis is consistent with the 2017 International Standards of Care for SMA (Mercuri et al., 2018). It should be noted that the role of genetic counsellors in SMA has adapted in the new therapeutic era (Serra-Juhe et al., 2019). Clinical geneticists and genetic counsellors will play important roles in collaboration with neurology specialists in terms of providing information around treatment options and timing, how treatment will be delivered and follow-up of patients. Additionally, at the appropriate time, information and advice surrounding future reproductive options can be discussed. • Recommendation 11.11 – comment on treatment options for infants with 4 SMN2 copies As outlined on pg 200 of the Guidelines document, at the time of writing, pre-symptomatic children with 4 or more *SMN2* copies do not have access to approved	These excellent points have been incorporated into Involving and acknowledging Aboriginal, Torres Strait Islander, Pacific Islander and Maori peoples and culturally and linguistically diverse communities. These excellent points have been incorporated into the Guideline on the expanding role of genetic counsellors. This now reads: With their role expanding in a new therapeutic era, genetic counsellors can now provide information not only on the genetics of a condition but work in conjunction with neurology specialists to facilitate understanding of treatment timing, delivery and follow-up. Dependant on health expertise and confidence in disclosing sensitive results to families, other programs have leveraged the experience of trained genetic counsellors or nurses, particularly in regional and remote areas.	

	and reimbursed treatments. This contrasts with an international consensus treatment algorithm (Glascock et al., 2020) which was inclusive of such infants. We note pt 4 of the 'Evidence gaps and future directions' relates to the management of newborns with SMA and 4 or more *SMN2* copies and the need for an increased evidence base for informed decisions regarding the risks and benefits of early treatment.	No changes required.	
	<u>Potential Guideline Impact</u> • Comment on likelihood of workforce issues for neurologists, GPs, genetic counsellors, laboratory diagnostic staff. In Queensland, an SMA newborn screening program has been in operation since May 2023 and it is anticipated that 6 individuals a year would be identified by the program, on average. Based on 2022 figures (D'Silva et al., 2022) and 300,000 births per year in Australia, one would expect 26-30 individuals per year affected by SMA. Given the complex nature of a multidisciplinary approach, workforce issues could be a barrier to successful implementation (as outlined on pg 198 of the National Guidelines). To mitigate such barriers, education of diagnostic laboratory workforce in terms of importance of turn-around-times for *SMN1* confirmation and *SMN2* copy number determination will be important. Regarding training, page 161 notes: "Non-specialist medical practitioners who may reasonably be expected to perform result disclosure where appropriate may require a process of training and education on SMA and implications of a screen positive result for optimal information provision". This may include Indigenous **Health Liaison Professionals** (IHLPs) but potentially other professionals in the Indigenous health workforce.	A sentence has been added to incorporate Indigenous Health professionals within an education and training model, within the future directions section; education and training for relevant medical practitioners in rural and regional areas.	
	<u>Overall feedback</u> We strongly support the proposal for guidelines to be **flexible** (pg 24, pg 25) which aligns with existing guidelines including the National Screening Framework and internationally developed Standards of Care for SMA. This is particularly relevant giving the likely ongoing advancements in treatment for SMA. We also support the proposed strategies for Guideline evaluation (pg 206/207) including the need for update of guidelines in a rapidly evolving landscapes, further investigation of barriers and enablers to implementation and acknowledgment of jurisdictional differences in adoption of the guidelines. In terms of the length of time for review – five years is suggested. This timeline seems appropriate; however, we envisage that any major changes in treatment or diagnostic methods may warrant an out-of-session review. As these are the first implementation of the guidelines, a 1-year 'fit-for-purpose' review could be of benefit. This would allow for adjustments based on any feedback from those	The need for a flexible approach to review of document is noted in the Future directions section which now reads: The Guideline should be reviewed (at maximum) in 5 years of publications or sooner if the screening, diagnostic or clinical landscape changes in the interim, updated to reflect and respond to new evidence from research, clinical practice and changes in community needs, values and preferences.	

	stakeholders who are utilising the guideline or identify any key gaps that might have only been highlighted once the guideline was used in the practical sense. We note that the 2016 NHMRC standards for guidelines state in section 6.1: Be informed by well conducted systematic reviews, however a timeframe is not given. <u>Broader feedback on relationship between NBS and RCS.</u> Pg 114 of the guidelines references the inclusion of SMA1 (and fragile X and cystic fibrosis) as a condition screened via reproductive carrier screening (RCS) (Medicare item number 73451). This will allow couples more information regarding their reproductive decision making in the context of SMA. The guideline document indicates the complementation of the two programs – this may warrant further comment and linking to guidelines for reproductive carrier screening as they become available. Potential bi-directional impacts of reproductive and newborn screening programs for certain conditions may include cost effectiveness, and awareness and education of the different health practitioners, including the strengths and limitations of screening programs in identifying conditions like SMA. <u>Possibility of generally streamlining Guidelines.</u> Due to the structured nature of their development there is some overlap between specific guidelines and the opportunity of streamlining. As an example, recommendation 8.4 and 8.5 concerning diagnostic results disclosure. We suggest such streamlining could be incorporated into future reviews. <u>Recommendation 11.5</u> We are very supportive of Recommendation 11.5 and the collection of real-world evidence by neurology services after identification and management of children identified as screen positive Post implementation evaluation metrics will be important to inform future refinement of the guidelines / screening practice. <u>Aboriginal and Torres Strait Islander, Pacific Islander and/or Māori representation on the GDG.</u> It was indicated that there was no formal representation of Indigenous populations on the GDG. We suggest invitation of consultation by respective groups such as Queensland Aboriginal and Islander Health Council (QAIHC), National Aboriginal Community Controlled Health Organisation (NACCHO), Te Aka Whai Ora (Māori Health Authority). This also relates to Recommendation 7.4 (pg 48). With no formal involvement, there was no clear messaging or guidance on how the lack of representation would be addressed within the framework. The	Whilst the SAC felt that comment on reproductive genetic testing was outside the scope of the current Guideline, the existence of guidelines for other screening methods for SMA was delineated in the Scope, population and setting section: Newborn screening is a public health program that fits alongside and within other public health initiatives such as reproductive carrier testing, and prenatal genetic screening. This Guideline acknowledges, compliments and does not replace existing guidelines that encompass these domains. As this is the first Guideline for NBS for SMA, we have adopted a structured approach and agree that the streamlining of recommendations can be considered at next review. No changes We agree with the stakeholder perspectives that these communities should be represented in future work. We have incorporated the advice for a Indigenous Advisory Group to support future research within the future directions page.	

	guidelines lay the responsibility for supporting families whose child has been diagnosed with SMA with the **Indigenous Health Liaison Professionals** to provide advice and be involved in how the clinical test is communicated to the family. This puts pressure on these roles/people and there are no clear recommendations for appropriate training that the IHLPs could be supported to undertake. Pg 210 refers to continued involvement of Aboriginal and Torres Strait Islander peoples in the evolving SMA research but no clear pathways identified for how this can be or should be achieved. In their current form the guidelines do not identify culturally appropriate pathways or best practice approaches to supporting Aboriginal and Torres Strait Islander families whose child has been diagnosed with SMA. We encourage the development of an Indigenous Governance Advisory Group to support ongoing guideline work.		

[redacted]	Technical report General comment As a general comment, the technical and administrative report was very useful, particularly the evidence tables for each section, for each respective recommendation. This will be a valuable resource for future revisions of the guidelines as the evidence base changes (for example relevant literature). Family fact sheet comments • The family fact sheet is an important communications tool and so Australian Genomics' community engagement team provide specific feedback to this section. This includes brief background on SMA, the guidelines process, a summary of screening, diagnostic and clinical care steps and a summary of recommendations. We suggest a further heading in slide 7 such as "Summary of screening and clinical pathway". • We also suggest mention (and link) to the Family fact sheet in the main Guidelines Document. What is SMA • Formatting of question mark at top and bottom • Instead of numbering each of the points, it may be better to use icons here that represent the content (e.g. a picture of someone walking/moving for point 2) • The gradient background could make it difficult for people who are vision impaired • More detail on inheritance may be warranted, for example, the sliders depicting percentage is a bit difficult to understand could use a pie chart or similar	-No change needed This title has been added. Family fact sheet now incorporated into main documents via link in the targeted secondary end users section. This has been changed Icons have been added Backgrounds have been placed in monotone for readability Changed sliders to pie charts. Added sentence "If both parents carry the gene mutation" to make clearer the linkage with % likelihood that child develops SMA. The wording has been changed and now reads, 'more copy numbers of SMN2' Order of circles changed according to feedback	

	• Great explainer of the cause of SMA but there is a new term "higher copy number" introduced at the end and not explained <u>What is NBS for SMA</u> • suggest changing the order of the circles – leading with what NBS is: 1. NBS aims to identify children at risk 2. This test takes a small amount of blood 3. NBS is offered to all babies 4. In Australia and NZ each health area 5. In 2022 and 2023 6. this is the first times genetic 7. Those identified during screening • Rather than "confirmatory testing" suggest " …urgently referred to confirm the results." • Formatting: Breaking up the heading at the top and bottom of the page make it difficult to read. <u>Why we need a guideline</u> • Content: The opening sentence "the intent of these guidelines…" is quite formal. Could reword to something like "These guidelines aim to provide recommendations that improve the care of newborns based on the best available evidence." • Formatting: Suggest placing text in boxes around the graphic <u>Steps page</u> • Content: 1. Steps could be reworded to the active voice e.g. Step 1 could be reworded to 'A dried blood spot is collected from the newborn for newborn screening'. 2. Step 2: Suggest "laboratory" rather than "reference screening" 3. Step 3: suggest removing "reference screening" and use laboratory. Spelling error: services. Could removing "screen" and replace with "positive result"	Words changed to match suggestion Heading from bottom brought under heading at top Words changed to match suggestion The SAC felt that this formatting change did not improve readability. Words changed to match suggestion Bold added to icons to ensure they are visible Co-leads feel Exon 7 is important in this context. The wording has been changed to make this style more in reflection of recommendations, linked in part to explanations to provide context. Gradient changed to single colour background We have reached out to the peak bodies for further consultation and have added the need for an Indigenous Advisory Group to inform further research. This now reads: the establishment of an Indigenous Advisory Group to inform future revisions and implementation of the Guideline will be a necessary future step towards equitable delivery of best care for all children with SMA across the diverse communities of Australasia.	

	4. Step 5: Suggest simpler explanation of "diagnostic evaluation". Spelling error: positive 5. Step 6: Suggest changing biomarkers to markers/signs. 6. Step 7: Reword 'The family is told the results and treatment plan starts' 7. Step 8: suggest rewording • Formatting: Icons are difficult to see. Would also make the outline of icons bolder <u>Summary page</u> • Screening box: Is there a need to mention exon 7? This has not been introduced previously. • Consider rewording of some of the Recommendations boxes, as some appear more to be explanations, rather than a summary of key recommendations. • gradient background will make it difficult for people who are vision impaired <u>Further general comments</u> [redacted] endorses the National Recommendations for Newborn Screening in Spinal Muscular Atrophy in Australia and New Zealand. Specific points of consideration: • Further engagement with Indigenous Health representatives and peak bodies across Australia and New Zealand. As stated previously, we suggest development of an appropriate Indigenous Governance Advisory Group to support this work. • Commend recommendations that address the **potential health inequity of access** to specialist neurology services and multi-disciplinary teams in outer regional, remote and very remote areas of Australia and New Zealand.	No change required. We have updated the need for a minimum 5 year review as above. No changes required. An implementation document has been provided as a separate file and is located on the website, with a link provided in the Guideline document under the section of future directions; dissemination and implementation of recommendations within the Guideline. This has been a point considered across the feedback. In response, the SAC agrees to add an implementation point in 10.1.1 that states: in Australia and Aotearoa New Zealand treatments for SMA are subsided by the publicly funded healthcare system for children who meet eligibility criteria. Reimbursement strudtures and options for treatment vary across the two countries. For children who are not eligible to access subsided treatments on the basis of their residency status or other factors, treatment pathways require interrogation on a case-by-case basis. The variations in practice and access to treatments have been added to the implementation point 10.1.1 which now reads In Australia and Aotearoa New Zealand treatments for SMA are subsidised by the publicly funded healthcare system for children who meet eligibility criteria. Reimbursement structures and options for treatment vary across the two countries. For children who are not eligible to access subsidised treatments, on the basis of residency status or other factors, treatment pathways require interrogation on a case-by-case basis. -No change needed	

	• We commend the need for **flexibility** in the guidelines given potential advancements in treatment and potentially developments in diagnostic technology. We suggest the possibility of out-of-session updates aside from the scheduled 5 years schedule for any major disruptive changes in treatment or diagnosis relating to SMA and newborn screening. • We agree with the section on pg 8 regarding **evidence gaps and future directions for stakeholders**. In relation to point 1- the evolution of genomics capabilities in newborn screening, we encourage further work in this area in benchmarking various platforms including exome and whole genome sequencing. Point 2 is also a very important consideration given the challenges in determining *SMN2* copy number and variables in linking copy number to disease prediction. • **Relationship and potential overlap between Guidelines and Implementation.** We note that there is considerable reference to downstream clinical management associated healthcare support that are very specific, given these are guidelines. It is not clear if a separate implementation document is planned at a separate stage. • **Although not directly addressed in the guidelines, individuals residing in Australia who are not eligible for Medicare do not have the same access to newborn screening or potential treatments.** We understand reimbursement of treatment in this scenario would be reviewed on a case-by-case basis on compassionate grounds which exacerbates inequities and widens the health gap. • There are a few differences between the Australian and New Zealand health systems relevant to SMA which may impact the guidelines – for example New Zealand currently funds Nusinersen as a treatment option, from January 2023 via Pharmac, New Zealand's pharmaceutical management agency (Pharmac 2022). Risdiplam was available from May 2023.		

	• **we reinforce the potential need for revisions of the guidelines,** given most of the evidence was consensus based. This may be particularly relevant for SMA given the rapid recent advancements in treatment and technologies relating to methodology.		

[redacted]	• Equity / rural and remote context Stakeholders uniformly highlighted that timely access to treatment services and teams may not be achievable in context of the timeframes recommended. The geographical size of [redacted] can present challenges for families in a rural or remote setting; their ability to access services and/or receive care in a timely manner is likely to be extremely challenging when considering the recommendations. Medicare eligibility of diagnosed infants can impact the ability to access specialist services. Confirmation testing of SMA is only available [redacted] and presents significant risk and delay to diagnosis and care of [redacted] infants. For rural and remote infants and their families, several stakeholders proposed that an adjustment to recommendations should be made to promote the increase of utilisation telehealth and local clinicians in an effort to reduce the impact on the centralised service and improve equity of access and support.	Whilst the SAC acknowledged the timelines for screening and diagnostic results could vary across health jurisdictions, due to the neurogenetic emergency of SMA, it was considered on the whole feasible to implement these timelines. Specific recommendations have been developed to help promote equity of access to best care for children in remote and rural areas. These include. Recommendation 8.5 Consensus based recommendation We suggest that if circumstances dictate and dependent on individual (family and child related) factors, it is acceptable for diagnostic disclosure to occur through telephone or Telehealth*. Grade of recommendation Conditional, Grade 2C *Child and family factors include but are not limited to geographical location, safety of travel for the child (relevant in a child with signs and symptoms of SMA), need for cultural or linguistic support to facilitate disclosure of the diagnosis and the availability of technology and connectivity for the use of telehealth. Section 5: Options include immediate referral to the neuromuscular team or, for those with difficulties travelling long distances, with the local paediatrician and specialist support using videoconferencing (telehealth) systems. Good Practice point 5.2.1. The process for communication of a screen positive result to families may be conducted through a telephone call or a telehealth consultation, and considers (if known), the families' comfort, convenience, privacy as well as practical considerations such as location and in the case of telehealth, access to appropriate and reliable equipment and connectivity. This is included in the implementation plan (linked to the Guideline) This is included in the implementation plan (linked to the Guideline) This is included in the implementation plan (linked to the Guideline)	

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		This is included in the implementation plan (linked to the Guideline)	
	• Workforce [redacted] noted that specialised allied health services were identified as a need, however, additional capacity in nursing and medical may be required to maintain or increase service provision based on the recommendations. Particularly, specialist neuromuscular clinicians are indicated to have key roles within the recommendations, however, the availability to resource this is not realistic in terms of clinical workforce availability and funding to resource services to the levels indicated in the recommendations • Service funding Funding for pre-screening and post-screening services does not specifically exist for newborn bloodspot screening. For post-screening services this especially presents a challenge when considering implementing the recommendations as essentially more services are being required without additional funding and resourcing to support them. • Service capacity Clinical and genetic services are currently operating at or over service capacity. If implemented, some of the recommendations will result in additional service delivery challenges to meet increased testing, family support, treatment, education, travel, and other needs. • First Nations		

	[redacted] emphasised that implementation of recommendations should include ensuring culturally appropriate and safe support for First Nations families with infants diagnosed with SMA. • Education Clinical education was highlighted as an essential component when considering implementation of the recommendations. Contemporary education for clinicians involved in pre and post-natal conversations, diagnosis, treatment and care of infants with SMA will strengthen their ability to provide safe, informed care.		

[redacted]	In principle agree with all recommendations; they are mostly consistent with the model-of-care in the neuromuscular service[redacted]. However, in order to continue to meet the recommendations there are some hurdles. • Equity a. [redacted] provides NBS for [redacted] – timely access to services and teams, may not be able to meet the timeframes recommended. [redacted] can offer telehealth for the initial conversation; however, these infants need some specific genetic and investigative blood sampling – this would be messy across health systems – challenging enough in [redacted]. Also, they need clinical examination by a Neurologist and physiotherapist who are specialists in SMA. The family would need to travel to [redacted], on short notice, within 1-2 days after NBS positive. Consideration for post-partum mothers and families is relevant given the geography of [redacted].	Whilst the SAC acknowledged the timelines for screening and diagnostic results could vary across health jurisdictions, due to the neurogenetic emergency of SMA, it was considered on the whole feasible to implement these timelines. Specific recommendations have been developed to help promote equity of access to best care for children in remote and rural areas. These include. Recommendation 8.5 Consensus based recommendation We suggest that if circumstances dictate and dependent on individual (family and child related) factors, it is acceptable for diagnostic disclosure to occur through telephone or Telehealth*. Grade of recommendation Conditional, Grade 2C *Child and family factors include but are not limited to geographical location, safety of travel for the child (relevant in a child with signs and symptoms of SMA), need for cultural or linguistic support to facilitate disclosure of the diagnosis and the availability of technology and connectivity for the use of telehealth. Section 5: Options include immediate referral to the neuromuscular team or, for those with difficulties travelling long distances, with the local paediatrician and specialist support using videoconferencing (telehealth) systems. Good Practice point 5.2.1. The process for communication of a screen positive result to families may be conducted through a telephone call or a telehealth consultation, and considers (if known), the families' comfort, convenience, privacy as well as practical considerations such as location and in the case of telehealth, access to appropriate and reliable equipment and connectivity. -This has been acknowledged in the implementation plan	

	b. Tier 2 genetic testing – In [redacted], confirmatory genetic testing needs to be sent interstate – [redacted] for SMN1 & SMN2 testing. Most states can offer this testing locally with a quicker turnaround time. At best these test results take 7-10 days for [redacted] families. This testing is essential to determine eligibility for PBS funded SMA medications. Testing and results is time critical.	The SAC acknowledges this point but felt it was outside the scope of the Guideline to address. This was added as a point in Scope, which now reads "It is made to be flexible and adapted to conform with available resources and capacity on a state/region/territory level across Australia and Aotearoa New Zealand. As such, it has been developed within the current health policy framework of these two countries and the parameters of the Guideline do not specifically address reimbursement pathways for children with SMA (diagnosed through newborn screening) who are not eligible for subsidised or publicly funded health services or treatments."	
	c. Medicare Eligibility – 50% of our patients diagnosed through NBS in [redacted] in 2024 have not had Medicare …. this impacts their ability to self-fund/access specialist NM services, allied health teams, and PBS funded treatments. They're also ineligible for NDIS. One family did not have private health insurance, which impacts delivering on Standards of Care (SOC) recommendations. The family does not have capacity to fund the appropriate standard of care.	-considered in implementation plan The GDG agreed that processes for result disclosures were jurisdictionally dependent, and that medical practitioners such as **genetic counsellors** nurse specialists and non-specialist medical practitioners could also be well placed to disclose and counsel on the results. For these professionals, the evidence showed that access to and advice from specialist services, enabled a streamlined and effective disclosure process.	
	d. Delivering care to SMA patients has impacted the NM service significantly with no additional resourcing. There are less appointments	-See implementation plan.	
	• Specialist nursing support a. Allied health teams were noted. Clinical nurse consultants/ nurse specialists weren't specifically mentioned, however have a vital role in supporting families from screen positive, through to coordination of care, clinical advice and ongoing specialist	-This is beyond the scope of this Guideline.	
	• Resourcing / funding of NM services / access to timely care a. NBS laboratory received funding to build capacity and capability of their service, however clinical services have not had additional funding to support care and management.	-Issues around resourcing are acknowledged in the implementation plan.	
	b. Psychological support for NBS positive – none at [redacted]. Our service has access to a Social Worker (SW) only, and we link all families with SW, however they also have other workloads and competing clinical commitments with other teams/inpatients etc. There is also a high turnover in the SW service for Neurology, so I would advocate for a consistent team that can develop specialised knowledge in this area. The SW do an excellent job; however, the turnover of staff is less than ideal. It's difficult for them to provide psychological support if they're only in the role for a few months.		
	c. Sustainability of services – Some states were successful in securing additional government funding. Unfortunately, our department, has absorbed		

	the NBS workload and treatments for SMA, however this has been challenging and workloads have increased significantly. Previously, palliation was the only option for many infants born with SMA, however they are now surviving, require high-cost PBS funded medications and intensive monitoring and coordination of care. We have had a 320% increase in SMA1 since 2018 when treatments became available. This means, higher number of patients, increased complexity and acuity. If we are to consistently deliver on the SOC recommendations it will be a challenge, without impacting other aspects of the service delivery in the Neuromuscular service and patients with other neuromuscular conditions. We are a very small team, resourcing and succession planning needs to be addressed. Services need to be reviewed and resourced accordingly. We already have long wait times for CAT 2 and review appointments. Timely access to ongoing care is a challenge, clinics are overbooked, and if a patient FTAs or cancels it's a 9 month wait for a review appointment. Currently all NBS SMA and SMA treatment monitoring are done over and above other workload. Appointments are booked adhoc and overbooked. This is not a sustainable system for patients or staff. Services cannot deliver the SOC recommendations without reviewing resourcing. d. SMN2 4 copies – impact on clinical services... frequency of reviews to monitor for disease progression, puts more demand on existing appointment availability. We know firsthand as we are one of the few states with an SMN2 4 copy patient. This patient became symptomatic … and was then eligible for PBS funded treatment. So close monitoring is very important to ensure timely initiation of treatment which can change long term health outcomes. e. Workforce for NM clinics – our service is significantly oversubscribed for appointments; we've had a reduction in medical FTE attached to the service and do not have capacity to absorb the workloads. Patients diagnosed with SMA need to be seen in a specialist NM service, however managing the demand and capacity is at a tipping point. We have done extensive work to ensure optimisation of services over the last few years, yet still struggle to see patients in clinically recommended time frames.		
[redacted]	1. Consensus based recommendation 7.4 on page 48 of the National Recommendations for Newborn Screening in SMA states "We recommend that medical practitioners providing information to, and discussing diagnosis with, families of newborns from Aboriginal, Torres Strait Islander, Pacific Islander, Māori or other First Nations backgrounds should be aware of particular issues arising from information provision and diagnostic evaluation. The medical practitioner may elicit the advice of Indigenous Health Liaison professionals in how to best conduct these evaluations and offer families the support of Indigenous Health Liaison services at the time of diagnosis." When considering appropriate support for First Nations families, consideration should be given to providing additional cultural support and sensitivity. We suggest a First Nations Nurse, Midwife or a Health Worker practitioner with a sound understanding of	- The suggested professionals have been incorporated into recommendation 5.5	

	the Newborn Screening process be included in conversations with these families where possible. 2. Consideration should be given to providing some detail about potential sensitivities for First Nations patients. This is not to remove the need for an Indigenous Health Liaison Officer or a First Nations health professional, but to provide better guidance for the clinician's discussions and to benefit the pursuit of cultural safety in the long-term with better understanding of this issues.	-Currently there is a paucity of evidence for potential sensitivities for First Nations peoples within the remits of NBS for SMA, as considered by a targeted systematic literature review. We have aligned our future directions to incorporate the need for research to address these data gaps.	
[redacted]	1. Supportive of the DRAFT Guideline supplied. 2. Makes perfect sense that the NBS recommendations align with current evidence base given treatment advancements for SMA. 3. The biggest factor for the midwifery cohort will not be the resources in terms of education and access to expertise for post diagnostic assessments but more so the educational requirements for having discussions with parents postnatally while gaining informed consent for NBS (with SMA screening included). 4. With the addition of SMA in the NBS will there be communication and educational update provided to maternity clinicians working with families at the point of NBS screening?	No change required. See implementation plan linked to the Guideline See implementation plan linked to the Guideline	
[redacted]	1. Agree with the draft documents rationale for including SMA testing, as described, in the routine NBS paradigm. 2. Recommendation 1.6 is important (not reporting heterozygous state) – reporting of carrier state would have significant implications for genetics services given the population carrier frequency for SMA. 3. Important to emphasise that inclusion of SMA on newborn screening will increase demands on neurology and clinical genetics services. Consequently, recommendations should also be made that Hospital and Health Services should ensure these clinical teams are appropriately resourced to meet the assessment / counselling demands that will result. 4. While those with clinical SMA would have been seen eventually by these services anyway, there is likely to be a false positive load that will increase work for both services. Given the nature of the condition, these families are still likely to need robust and timely counselling	No change required No change required Resourcing found in the implementation plan linked to the Guideline.	
[redacted]	•Consensus feedback 1. The document is comprehensive however at over 200 pages it may impact readability.	Whilst we acknowledge that the Guideline is 200 pages, it provides a comprehensive view of the separate domains of screening, diagnosis and clinical care. Whilst it can be read from top to bottom, for ease of reference, readers can go to individual sections easily.	

[redacted]	2. There are many repetitive statements, with the formatting impacting on the ease of reading the document. 3. The suggested requirement for the availability of a paediatric neurologist as the point of contact and the person for initial screening mentioned throughout may be impractical, especially [redacted]. For reference, there is one paediatric neurologist [redacted], but otherwise no others outside the [redacted]. Relying on the sole practitioner for a very large area to be available may be a quite cumbersome and risk delays in diagnoses. Currently, [redacted] has an effective system for following up abnormal results, involving the appropriate teams from Metabolic, Immunology or Neurology, in which the results then defer to the local delivery/paediatric centre. This works well for metabolic conditions which require very rapid management. [redacted] suggests utilising the already well-established system, along with a co-referral to the paediatric neurologist as a consideration. – a query for [redacted] is, will the neurologist at [redacted] be deemed the link person for the state?	The emergency nature of SMA warrants specialist input and therefore the SAC maintains that a paediatric neurologist should be contacted for the screen positive result. [redacted] were part of the consultation process and have agreed to this recommendation. We acknowledge that work flow will vary between health jurisdictions and this has been accounted for in a slight rewording of these recommendations as follows: Recommendation 2.7. Consensus based recommendation We recommend that the newborn screening for SMA program will establish a clinical referral pathway for newborns who screen positive for SMA. A positive newborn screening result should be verbally relayed to a designated paediatric neurologist. Grade of recommendation Strong, Grade 1C Implementation point 2.7.1. In many health jurisdictions, newborn screening programs have an established notification strategy that involves notifying a healthcare practitioner (usually general practitioner, obstetrician, maternity nurse or paediatrician) on the child's dried blood spot demographics. Due to the imperative to have access to expedient diagnosis and treatment, newborn screening programs should establish a clinical referral pathway that includes simultaneous, early notification of a screen positive result to a paediatric neurology specialist. Good Practice point 2.7.1. We suggest that the newborn screening for SMA program will establish a clinical referral pathway for newborns who screen positive for SMA. A positive newborn screening result may also be verbally relayed to a relevant listed healthcare practitioner (that is or has been involved in the care and management of the child such as a general practitioner or paediatrician). This is the English/Australia spelling of foetal and therefore has been retained. Extra space entered between sentences This has been changed to read designated healthcare practitioner. This has been added to the recommendation	

	4. Page 104 – there is a spelling error, foetal should be corrected to- fetal. • Additional late feedback 1. Page 33 Recommendation 2.7 – Formatting error – needs a space inserted between the first two sentences, highlighted in yellow, for readability - We recommend that the newborn screening for SMA program will establish a clinical referral pathway for newborns who screen positive for SMA.A positive newborn screening result should be verbally relayed to a designated paediatric neurologist. 2. I agree group, the pathway including the handling of false positive results, should follow that already established for NBS. 3. Page 42 Recommendation 5.3 – this wording could be changed to 'responsible healthcare practitioner' instead of medical. For example, a specialist neurology nurse practitioner or genetic counsellor with support from a paediatric neurologist would be a suitable person to disclose a screen positive result, the latter not typically falling under the descriptor of 'medical' which could be taken to mean doctors only, or doctors/nurses but would typically not be used as a descriptor of allied health including genetic counsellors, who are arguably well placed to perform this role. This would also make this recommendation congruent with the following recommendation 5.4, which does reference healthcare practitioners. 4. Page 48 Recommendation 7.5 – should include clinical geneticist or genetic counsellor or genetic service. 5. Page 106 – the use of the term 'healthy individual' is not in line with best practice around the language used in disability, as it's a value laden term that many parents of children with a disability find distressing. Alternative terminology has been recommended. Equally, 'SMA patient' is better stated as Individual with SMA or Neonate with SMA or Child with SMA or Person with SMA. Recommend "person without SMA" and "person with SMA" for this section.	-Changed 'healthy individuals' to 'individuals without SMA'. -All mentions of 'patient' are in the context of definitions by CLSI, or 'patient organisation' etc.	
[redacted]	1. There should be more consideration/emphasis for patients and families who live in more rural/remote regions of the country (e.g. rural QLD and WA, the NT) who are already at a disadvantage from receiving high quality healthcare. Most families from rural QLD and WA, as well as the NT are often more than a couple hours away from their local tertiary paediatric hospital.	The SAC agrees with these comments and have accounted for this in the recommendations as follows, with wording changed to incorporate the wider role of general practitioners. Grade of recommendation Conditional, Grade 2C	Some adjustments made to account for the feedback but the recommendati

	a. Travel with a young infant, especially when they are initially diagnosed can often be challenging. b. The utilisation of telehealth and local medical resources might be an avenue to emphasise and consider. c. For example, at the initial consultation/on initial diagnosis, patient and family can be with the local paediatrician, and the paediatric neurologist can provide the initial consult via telehealth. Additionally, this method can be used to support the local paediatric team during subsequent reviews. 2. Should there be more involvement of a general paediatrician in the holistic care of these children, especially ones who live in rural/remote regions of the country, where access to a specialist multidisciplinary clinic might be challenging to access. a. Involvement of a local general paediatrician, especially at the time of diagnosis, gives these patients a local contact person, but also someone who can coordinate the patient's overall care (e.g. growth, development etc).	*Child and family factors include but are not limited to geographical location, safety of travel for the child (relevant in a child with signs and symptoms of SMA), need for cultural or linguistic support to facilitate disclosure of the diagnosis and the availability of technology and connectivity for the use of telehealth. Section 5: Options include immediate referral to the neuromuscular team or, for those with difficulties travelling long distances, with the local paediatrician and specialist support using videoconferencing (telehealth) systems. Recommendation 5.2 Consensus based recommendation We recommend that the designated paediatric neurologist, receiving the screen positive SMA result (recommendation 2.10), should coordinate with other relevant healthcare practitioners to develop a family-centred plan for screen positive disclosure, including delegation of roles for who is best placed to facilitate this process. Grade of recommendation Strong, Grade 1C Good Practice point 5.2.1. The process for communication of a screen positive result to families may be conducted through a telephone call or a telehealth consultation, and considers (if known), the families' comfort, convenience, privacy as well as practical considerations such as location and in the case of telehealth, access to appropriate and reliable equipment and connectivity. Recommendation 5.3 Consensus based recommendation We suggest that it is acceptable for a designated healthcare practitioner with support from a paediatric neurologist to disclose a screen positive result to a family. Grade of recommendation Conditional, Grade 2C *The designated healthcare practitioner will vary between health jurisdictions and may include general practitioners, paediatricians, specialist nurses and/or genetic counsellors. These individuals should have training and expertise in disclosing screen positive SMA results to families.* *Support as defined in this recommendation can range from exchange of advice, information (verbal and/or written) or a formal offer to be part of the screen positive disclosure, alongside the designated healthcare practitioner.* *Recommendation 5.8* *Consensus based recommendation* *We suggest that a clinical review within local paediatric services, with clinical support from a paediatric neurologist should be offered to screen positive newborns where access to specialist (neurology) services is limited and may cause delay in diagnostic evaluation.* *Grade of recommendation Conditional, Grade 2C* *The local healthcare practitioner will vary between health jurisdictions and may include general practitioners, paediatricians and specialist nurses. These individuals should have training and expertise in the clinical evaluation of screen positive children with SMA.* *Support as defined in this recommendation can range from exchange of advice, information (verbal and/or written) or a formal offer to be part of the clinical review of the child, alongside the designated healthcare practitioner, using telehealth systems to augment this recommendation as appropriate.*	ons are across the board centred on equity of access in regional and rural areas.

		Recommendation 8.5 Consensus based recommendation We suggest that if circumstances dictate and dependent on individual (family and child related) factors, it is acceptable for diagnostic disclosure to occur through telephone or Telehealth*. *Child and family factors include but are not limited to geographical location, safety of travel for the child (relevant in a child with signs and symptoms of SMA), need for cultural or linguistic support to facilitate disclosure of the diagnosis and the availability of technology and connectivity for the use of telehealth. Recommendation 10.11 Consensus based recommendation We suggest that for some newborns, SMN augmenting treatments may be planned to be initiated from a non-specialist treatment centre, with paediatric neurology support.* Strength of recommendation Conditional, Grade 2C *This recommendation may be appropriate for children living in regional and rural areas where travel to paediatric neurology treatment centres is logistically or clinically challenging. Recommendation 10.12 Consensus based recommendation We suggest that post treatment monitoring for newborns who access SMN augmenting treatments may be shared between paediatric neurology centres, secondary paediatric services and community (general practitioner) services (with support from the specialist centres) as child and family factors dictate. Strength of recommendation Conditional, Grade 2C Recommendation 9.4 Consensus based recommendation We suggest that all children diagnosed with SMA through newborn screening should have a shared model of care between local community (general practitioners and allied health therapists), secondary (paediatric services) and specialist (paediatric neurology) services, to facilitate post diagnosis care, which is personalised according to the biopsychosocial characteristics of the child and family. Strength of recommendation Conditional, Grade 2C This is ideal but felt to be outside the scope of this document.	

	3. The guidelines should strongly encourage the development of a state based neuromuscular clinic (which I suspect is likely to be available in all tertiary paediatric hospitals across the country), where there can be multidisciplinary		

	review of these patients. Additionally, these clinics should also closely liaise to regional teams (including various allied health teams) to empower them to help provide care to these patients in rural and remote regions.		
[redacted]	It's great to see in the evidence gaps and future directions for stakeholders' section of the Guidelines, there is reference made to broadening and deepening the evidence base of perspectives and challenges for families from rural and remote regions. We would recommend that rural and remote families are prioritised for co-design of educational resources for families.	In the future directions section we have incorporated specific mention of rural populations and their role in future co-design. "Given the unique challenges facing rural and remote regions, it remains a priority to incorporate representative voices of this population into any future co-developed evidence. Furthermore, the information gap at the point of screening, diagnosis and therapeutic decision making for families can only be filled through codesign of targeted and relevant educational resources with the child and family perspective to remain central.	
[redacted]	1. [redacted] has no comment. The guideline appears comprehensive on the topic. Recommendations noted for inclusion in the [redacted] guideline (in development). 2. Note also that SMA forms part of the reproductive genetic carrier screening recommendations as per the [redacted]: Preconception and prenatal genetic screening clinical guideline.	-No change required. This has been addressed within the Scope section which now reads: Newborn screening is a public health program that fits alongside and within other public health initiatives such as reproductive carrier testing and prenatal genetic screening. This Guideline acknowledges, compliments, and does not replace existing guidelines that encompass these domains.	