Newborn screening for spinal muscular atrophy

Guidelines for Australia and New Zealand

National Recommendations for Newborn Screening in Spinal Muscular Atrophy in Australia and New Zealand

Feedback

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The Guideline

Feedback for screening

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

Evidence based recommendation

We recommend that the target analyte of newborn screening for SMA is homozygous deletion of exon 7 on *SMN1*.

Grade of recommendation B

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.

Recommendation 2.4. (pg 33, 130)

Consensus based recommendation

We recommend that the (in)availability of *SMN2* copy number should not delay clinical notification of a screen positive result based on absence of exon 7 on *SMN1* on newborn screening.

Grade of recommendation Strong, Grade 1C

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).

Feedback for diagnostics

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.

Recommendation 3.4 (pg 35, pg 140)

Consensus based recommendation

We suggest that diagnostic *SMN1* testing is conducted using a different methodology to the newborn screening assay.

Grade of recommendation Conditional, Grade 2C

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

Consensus based recommendation

We suggest that diagnostic test results (including *SMN1* and *SMN2* copy number) should be available to clinical services within 30 days of birth.

Grade of recommendation Conditional, Grade 2B

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

Consensus based recommendation

We suggest that diagnostic reports should detail the methodology used for analysis and the precise *SMN2* copy number (avoiding reports such as *SMN2* ≥4).

Grade of recommendation Conditional, Grade 2B

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.

Feedback for clinical care

Recommendation 5.3

Consensus based recommendation

We suggest that it is acceptable for a responsible medical practitioner with support from a paediatric neurologist to disclose a screen positive result to a family.

Grade of recommendation Conditional, Grade 2C

Recommendation 8.2.

Consensus based recommendation

We suggest that ideally, diagnostic results should be disclosed to families by a specialist medical practitioner such as a paediatric neurologist.

Grade of recommendation Conditional, Grade 2C

Recommendation 9.7

Consensus based recommendation

We suggest that for sibling(s) of affected children who live in remote regions, a review for signs and symptoms of SMA may be offered and conducted by a local medical practitioner, with support from a paediatric neurologist.

Strength of recommendation Conditional, Grade 2C

Recommendation 10.10

Consensus based recommendation

We recommend that the administration of SMN augmenting treatments should occur in a specialist (paediatric neurology) care centre.

Strength of recommendation Strong, Grade 1C

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 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

Consensus based recommendation

We suggest that families of newborns diagnosed with SMA through newborn screening programs should be offered referral to, and review at a clinical genetics service for genetic counselling and cascade screening.

Strength of recommendation Conditional, Grade 2C

Australian Genomics 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.

Comment on treatment options for infants with 4 *SMN2* copies (Recommendation 11.1).

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 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.

Potential Guideline impact

Barriers and facilitators of implementation of recommendations

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.

Feedback for the Guideline overall

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 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.

Broader feedback on relationship between NBS and RCS.

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.

Possibility of generally streamlining Guidelines.

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.

Recommendation 11.5 (pg 203)

Consensus based recommendation

We suggest that national clinical paediatric neurology centres should coordinate and establish databases to collect outcome data for newborns who have ≥ 4 *SMN2* copies and are under clinical surveillance, to establish an evidence-base to guide therapeutic and policy decision making.

Strength of recommendation Conditional, Grade 2C

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.

Aboriginal and Torres Strait Islander, Pacific Islander and/or Māori representation on the GDG.

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 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.

Technical and administrative report

Feedback

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 – brief overview

Feedback

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.

The following specific suggestions are made on a slide-by-slide basis:

What is SMA?

Formatting:

- 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

- Content:
  - 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
  - Great explainer of the cause of SMA but there is a new term "higher copy number" introduced at the end and not explained

What is NBS for SMA?

Content:

- suggest changing the order of the circles – leading with what NBS is
  1. NBS aims to identify children at risk
  3. This test takes a small amount of blood
  4. NBS is offered to all babies
  5. In Australia and NZ each health area
  6. In 2022 and 2023
  7. this is the first times genetic
  8. 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 is difficult to read

Why we need a guideline?

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

Steps page

Content:

- 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'.
- Step 2: Suggest "laboratory" rather than "reference screening"
- Step 3: suggest removing "reference screening" and use laboratory. Spelling error: services. Could removing "screen" and replace with "positive result"
- Step 5: Suggest simpler explanation of "diagnostic evaluation". Spelling error: positive
- Step 6: Suggest changing biomarkers to markers/signs.
- Step 7: Reword 'The family is told the results and treatment plan starts'
- Step 8: suggest rewording

- Formatting:
  - Icons are difficult to see. Would also make the outline of icons bolder

Summary page

Content:

- 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

Additional feedback

If you would like to leave additional feedback about anything else please do so here

Australian Genomics is an Australian Government initiative supporting genomic research and its translation into clinical practice. Australian Genomics supports Commonwealth, State and Territory health departments in the implementation of genomics research outcomes by refining and communicating evidence to inform policy development. Australian Genomics is a key supporter of the emerging Indigenous Genomics agenda, most visible through its direct support for the Australian Alliance for Indigenous Genomics (ALIGN), funded through the Medical Research Futures Fund (MRFF). ALIGN also contributed to this response.

Australian Genomics formed the Genomic Screening Consortium for Australian Newborns (GenSCAN), which includes the lead investigators of each of the five projects. GenSCAN was developed for the purpose of enabling improved efficiency and impact of the MRFF GHFM investment through complementary and collaborative research, as well as a cohesive national approach to the exploration of genomics into Australian newborn screening programs.

Australian Genomics 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.

- 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.

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