2025

Newborn Screening for Spinal Muscular Atrophy in Australia and Aotearoa New Zealand

Dissemination & Implementation Plan

Dissemination and implementation of the Guideline

Overview

This Guideline provides a set of Evidence and Consensus recommendations for newborn screening for SMA across Australia and Aotearoa New Zealand. As such it is relevant to all health jurisdictions undertaking newborn screening programs for SMA across Australasia. To ensure this is carried out equitably and efficiently, the dissemination and implementation of the Guideline is a necessary step to inform policy and practice and evaluating its usefulness and impact.

The Guideline should be reviewed and updated (at maximum) in 5 years (that is on or before the 1st of April 2030) or sooner if the screening, diagnostic or clinical landscape changes in the interim. The Guideline should be updated to reflect and respond to new evidence from research, clinical practice and changes in community needs, values and preferences. The methodology employed for the update should identify and prioritise topics required for the identification of a new evidence base published since the search period for the existing Guideline. A future revised Guideline should advise on the scope and clinical questions for the evaluation and methods to identify and evaluate relevant evidence. continue to be systematic and align with the recommendations and approvals required by the National Health and Medical Research Council.

Dissemination

Pursuant to the publication of the Guideline, dissemination will be facilitated primarily through the Organising Committee, and further facilitated through a range of activities, conducted in close liaison with relevant professional colleges, societies and consumer representative organisations (Table 1). It is planned that activities will include dissemination through the International Guideline Portal and the University of New South Wales who will house the Guideline and associated documents on a dedicated website (https://www.unsw.to/nbs-sma). Dissemination of the Guideline will also be in the form of promotion within newsletters, social media, websites, and utilisation in student teaching within the teaching hospitals across Australasia. To date, systematic reviews of available literature spanning the entire newborn screening for SMA journey are not part of the scholarly literature and thus it is envisaged that

manuscripts will be developed pertaining to the systematic literature review that formed the evidence base for the recommendations and published in a peer review journal.

Furthermore, emails will be delivered to organisations that have endorsed the Guideline, to members of the GDG for distribution to relevant stakeholders, to individuals or organisations providing feedback during the public consultation process and through national and international presentations to the scientific, clinical and SMA advocacy/consumer communities.

Table 1. Professional and consumer organisations invited to distribute the Guideline

Organisation	Audience
All state and federal health departments	Policy makers/ jurisdictional responsibility
Australian and New Zealand Child Neurology Society	Clinical decision making
Australian Genomics	Clinical decision making
Australian College of Rural and Remote Medicine	Clinical decision making
Australasian Association of Clinical Geneticists	Clinical decision making
Australasian Society of Diagnostic Genomics	Clinical decision making
Human Genetics Society of Australasia	Clinical decision making
Ministry of Health – Manatū Hauora	Policy makers/ jurisdictional responsibility
New Zealand Paediatric Society / The Paediatric Society of New Zealand	Clinical decision making
Queensland Aboriginal and Islander Health Council	Policy representatives, and advocates for Aboriginal health
Rare Disease Foundation Australia	Advocacy groups and families of children screened positive for SMA
Rare Disorders NZ	Advocacy groups and families of children screened positive for SMA
Rare Voices Australia	Advocacy groups and families of children screened positive for SMA
SMA Australia	Advocacy groups and families of children screened positive for SMA
Syndromes Without a Name	Advocacy groups
The National Aboriginal Community Control Health Organisation	Policy representatives, and advocates for Aboriginal health
The Royal Australian College of Physicians	Clinical decision making

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists	Clinical decision making
The Royal Australian College of General Practitioners	Clinical decision making

The methods of dissemination and purpose for each consumer group (healthcare practitioners, general public, consumer representatives, researchers, government sector) are discussed below (Table 2)

Table 2. Dissemination methods for stakeholder type

Audience	Purpose	Method
Healthcare practitioners	Increase awareness and adaptation of Guideline Improve best standards of care Ensure equitable and timely delivery of services	GDG to circulate with peers. Conference presentations. (e.g. ANZCNS congress, RACP congress, HGSA annual scientific meeting) Forwarded via organisations contacted (Table 1) Published in International Guidelines Network Publications in journals Incorporation into student teaching
Researchers	Increase awareness of Guideline Contribute to international best practice standards around newborn screening and SMA.	Conference presentations (e.g. ANZCNS congress, RACP congress, HGSA annual scientific meeting) Publications in journals Published in International Guidelines Network GDG to circulate to peers
Consumer representatives	Increase awareness of Guideline. Ensure advocacy is in line with best practice expectations.	Personalised emails to relevant representatives with links to documents. SCHN/UNSW media launch and external media engagement
Government sector	Increase awareness of Guideline Jurisdictional responsibility for ensuring standards are met.	Personalised emails to relevant representatives with links to documents.

General public	Increase awareness of Guideline	Newsletters and social media SCHN/UNSW media launch and external media engagement

Abbreviations: GDG, Guideline Development Group; ANZCNS, Australia and New Zealand Child Neurology Society; RACP, Royal Australasian College of Physicians; HGSA, Human Genetics Society of Australasia; SCHN, Sydney Children's Hospital Network; UNSW, University of New South Wales.

Evaluating the effectiveness of dissemination

The evaluation of the dissemination phase will be considered from the perspectives of healthcare practitioners, jurisdictional bodies, consumers and consumer representatives, and the public. Continual evaluation of the effectiveness of dissemination will be enabled through a dedicated section on the website for ongoing feedback and impact of the Guideline.

Table 3. Evaluation methods and metrics

Evaluation tool	Details	Proposed Metrics
Downloads	Total download number for each document via UNSW website	150 downloads within first 12 months
Website traffic	Total views for each document via UNSW website	1000 views within first 12 months
Conference presentations	Total number of presentations to target audiences	Guideline presented at 4 national conferences within 12 months and 2 international conferences within 12 months
Consumer surveys	General awareness of documents (in particular family fact sheets) for parents who have received an SMA positive result for their child	Metrics to be established by consumer advisory group (high level of awareness expected)
Healthcare surveys	General awareness of documents for relevant professionals.	Metrics to be established by consumer advisory group (high level of awareness expected)
Endorsements	Total number of organisations	Endorsements by 12 organisations, including primary targeted organisations (SMA advocacy groups, ANZCNS, HGSA).
Jurisdictional incorporation	Total number of health jurisdictions utilising the Guideline	All government bodies representing states, territories, and regions.
Social media	Total number of posts	12 posts by relevant organisations

Traditional media	Total number of articles	2 articles published
Scientific articles	Impact of journal article	Citations

Implementation

The overall goal of the Guideline is to standardise newborn screening for SMA to diagnose children and improve access to management for children with this condition, and to optimise their health and psychological benefits. The implementation of recommendations in the Guideline are the responsibility of each state and territory in Australia (which has a non-federated system) and of Aotearoa New Zealand. The implementation of the Guideline is facilitated by the fact that newborn screening programs are well established across Australasia and screening for SMA will be incorporated into routine screening panels. Scoping programs have been conducted in several jurisdictions to establish the barriers, facilitators of implementation and best practice standards.(1, 2)

The GDG acknowledge that workforce capacity varies across health jurisdictions and that implementation of the recommendations in the Guideline will require appropriate healthcare planning and resourcing to facilitate implementation and sustainability of services. These include health policy decisions on appropriate resourcing for screening and diagnostic purposes alongside allocation of provisions for meeting Guideline requirements within paediatric (specialist and non-specialist) services, genetic testing and counselling domains, and multidisciplinary healthcare services.

Whilst all recommendations in the Guideline are considered as **key recommendations** and as such should be implemented, consensus recommendations have associated prioritisation categories which are meant to help healthcare jurisdictions implement recommendations in a staged manner based on their priority level. However, the Guideline and the recommendations therein are an adjunct to and do not replace healthcare practitioner judgement in each case. Guideline More details on the barriers to implementation and the methods with which they may be mitigated are discussed in Table 4.

Table 4: Potential barriers/risks and their mitigating strategies to facilitate implementation

Barrier	Contextual factors	Mitigating strategies
Challenges in accessing healthcare practitioners to conduct post diagnosis activities, treatment planning, care and support in a timely manner.	Health care workforce capacity limited in some jurisdictions and typically located in specific (metropolitan) hubs.	Leveraging existing Australasian healthcare infrastructure that includes well established specialist (paediatric neurology), children's (paediatric) and multidisciplinary services. These networks are used to managing children within and between states and territories and accepting and prioritising referrals for children with emergency and complex needs. The Recommendations within the Guideline have been formed from an Australasian perspective with specific consideration regarding the need to work collaboratively within networks and prioritise children with a positive screening result.
Inability to identify all children at risk of SMA with current target analytes and assays.	Current screening assays identify the 95% of children with absence of *SMN1* due to biallelic deletion on exon 7.	Ongoing national/international research into new technologies that can identify the 5% of the population with an alternative genotype and future review of the Guideline to align with the changing landscape of genomic technologies.
Increased demand for reproductive counselling, cascade testing and preimplantation genetic testing	Altering the diagnostic pathway, shifting it from a clinical diagnosis triggered by clinical signs to a newborn screening triggered diagnosis.	Expansion of neurogenetic services and adopting the MDT style of care (with access to genetic and neurology services in one location). Members of the wider multidisciplinary team could augment roles as information and support providers dependent on jurisdictional resources and capacity. Updating educational resources to provide tailored and accurate information regarding the processes and implications of reproductive genetic testing.
Lack of speciality knowledge, and access to those with specialist knowledge.	Particularly noticeable for rural regions where specialists are less accessible.	Utilisation of telehealth services to enable a hub and spoke model of care where paediatric neurology services guide and support local healthcare practitioners in post diagnostic care and treatment surveillance.

		Education program development with implementation strategies to be codesigned with relevant stakeholders to disseminate knowledge of the condition, treatment options and best practice considerations.
Different organisational demands and infrastructure between health jurisdictions.	Laboratories may use different technology and protocols. Varying catchment areas for laboratories and hospitals may lead to different processing times.	Recommendations are assay agnostic and therefore there is flexibility for each jurisdiction to utilise technology and services available to them.
Widening of health inequities	Introduction of genetic testing to the newborn screening program may lead to disengagement and reduce the uptake for the overall program. Data sovereignty and potential for genetic discrimination may be particularly important concepts for indigenous families and those within CALD populations.	Public dissemination of information as to the benefits and risks of newborn (genetic) screening for SMA and educational resources that are codeveloped by these groups and address their specific needs and concerns.
Missing awareness about this Guideline and why it is necessary, by healthcare practitioners and advocacy groups.	Newborn Screening for SMA is a recent development and not all states and territories currently administer these pathways.	The Guideline will be disseminated across the spectrum of stakeholders through relevant channels (Table 1).
Missing awareness about this Guideline by families/ general public.	SMA is a rare disease and relatively unknown within the broader community. Best standards of care are similarly unknown.	Co-design and co-development of educational resources for families and advocacy groups guided by the formation of a National Consumer Advisory Group (CAG). The CAG will contribute to equitable access to information and support across Australasia, enabling the successful translation of the Guideline. This group will seek specific input

		from Culturally and Linguistically Diverse groups and Aboriginal, Torres Strait and Pacific Islander, and Māori people, and will be tasked with the responsibility of ensuring relevant platforms provide the necessary education nationally (including the production and dissemination of multimedia resources), while aligning with this Guideline. (Recommendations: 5.3,7.2, 7.3)
Inequitable access and delivery of healthcare arising from sociodemographic factors including cultural and linguistic barriers	Contributing factors include health literacy, socio-economic differences, geographical location of communities in relation to health services.	The CAG will seek specific input from Culturally and Linguistically Diverse groups including Aboriginal, Torres Strait and Pacific Islander, and Māori people to develop resources that are informed by and meet the needs of the community. Financial and travel support for families with financial difficulties to enable access to best care and treatment. Education and training for the Indigenous healthcare workforce on the aspects of newborn screening for SMA that can facilitate support for families (Recommendations 5.3. 7.3.)
Consistent application of the Guideline over time	Interest may peak at the initial implementation but peter out as activities lessen.	Screening and diagnostic laboratory annual reports will be part of quality assurance, auditing activities. Clinical services will be encouraged to audit post diagnosis activities, pathways and outcomes as part of quality improvement studies. The Guideline will be reviewed at maximum within 5 years to ensure it adequately meets best practice standards and those standards are being met. Auditing of screening and diagnostic services, along with clinical referrals and time of diagnosis, will reveal whether outcomes are consistent, and what alterations are necessary.
Challenges satisfying the timelines within the recommendations	Costs associated with personnel and staff time to expedite diagnostic assays and reporting Geographical challenges in specimen collection and	Where possible, recommendations take advantage of existing structures and processes within the Australasian healthcare system. Auditing of screening and diagnostic services and timelines will indicate where changes are necessary. (Newborn) screening laboratories have pre-established annual audits of

	distribution to diagnostic laboratories and access to specialist services for families for regional and rural communities. Timely access to necessary services	implementation timelines, and accuracy of assays which will be leveraged to facilitate streamlined processes and maintain the quality of newborn screening for SMA. Utilisation of technology to streamline processes and overcome geographical distances including telehealth., empowering to local healthcare practitioners to facilitate care and intervention (with paediatric neurologists supporting this process). Jurisdictions will be encouraged to establish a workflow that involves coordination and communication between screening, diagnostic and clinical care stakeholders to meet the timelines within the Guideline.
Costs of implementation	Healthcare resourcing is finite within Australasia with complex funding streams for screening, diagnosis and clinical care services.	Economic analysis shows that newborn screening for SMA coupled with treatment reduces long term costs and associated demands on healthcare services.

Evaluating the impact and implementation of the Guideline.

Key considerations will include but are not limited to, jurisdictionally dependent feasibility and sustainability of implementing the recommendations, effects on equity of access to diagnosis and care, effects on clinical practice and health system readiness for a change in workflow with the addition of SMA into routine newborn screening, and the short and long term clinical and psychosocial outcomes for children and their families. Systematic evaluation of the implementation and impact of the recommendations will thus facilitate wide stakeholder engagement to build resources, infrastructure and logistical capabilities to sustain an effective program of newborn screening for SMA into the future. The members of the organising committee have expertise in clinical research and implementation science and are well placed to evaluate the awareness, understanding and impact of the Guideline. As such, it is envisaged that the impact and implementation of the Guideline may be evaluated using the following strategies.

Table 5: Mixed qualitative and quantitative methods to assess awareness, understanding, and impact of the Guideline.

Evaluation strategy	Details
Longitudinal data collection of outcomes for newborns diagnosed with SMA	This will consider health indicators for newborns diagnosed with SMA with newborn screening. In particular, improvement in quality of life, attainment of motor milestones, and time to diagnosis within and between health jurisdictions. Particular attention will be given to comparisons of health outcomes between areas of high and low Guideline uptake.
Screening laboratory annual reports	Determine the timing and process of newborn screening for SMA. These assessments are conducted as part of formal quality assurance, and audit activities that evaluate newborn screening programs. (Recommendations: 1.1, 1.2, 1.3, 2.1, 2.2, 3.2, 4.1, 4.2, 5.1)
Evaluation of model of care across health jurisdiction	This may include assessment of the temporal processes such as time to screen positive result, diagnostic evaluation, confirmation of diagnosis and time to treatment plan and initiation alongside the longitudinal evaluation of the short- and long-term clinical outcomes for children screening positive for SMA. (Recommendations: 1.2, 1.3, 2.1, 2.2, 3.3, 3.4, 5.1, 5.2, 61, 8.4)
Consumer surveys for general public	The public acceptability of the newborn screening for SMA program as guided by the recommendations within the Guideline, and the barriers, facilitators and impact of implementation from a consumer perspective. (Recommendations 4.1, 5.3, 7.2, 8.5). These surveys will also seek to evaluate consumer understanding and knowledge of newborn screening for SMA.
Consumer surveys for CALD, Aboriginal, Torres Strait, Pacific Islander, & Māori peoples.	To ensure equitable delivery of healthcare to all Australians and New Zealanders, these surveys will be conducted to ensure accessibility, awareness, understanding, and use is felt and delivered equally to these communities, when compared with the general public. (Recommendations 5.3, 7.3)
Healthcare professional surveys	These surveys will evaluate whether the Guideline has changed clinical practice and the magnitude and direction of this change. This survey will seek to evaluate challenges arising for healthcare practitioners in screening, diagnostic, clinical care, and advocacy domains. Surveys will also be utilised during a maintenance phase to understand challenges that may arise if initial interest and awareness in the Guideline changes. Particular attention will be given to understanding why Guideline uptake may differ between regions and what can be done about this.

Sustainability and economic analysis	To determine capacity restraints, human resource availability, intervention costs, staff recruitment and turnover, and local context adaptation. This will be vital to the Guideline review process.

Abbreviations: CALD, Culturally and Linguistically Diverse

1. D'Silva AM, Kariyawasam DST, Best S, Wiley V, Farrar MA, Group NSNS. Integrating newborn screening for spinal muscular atrophy into health care systems: an Australian pilot programme. Dev Med Child Neurol. 2022;64(5):625-32.

2. Kariyawasam DST, Russell JS, Wiley V, Alexander IE, Farrar MA. The implementation of newborn screening for spinal muscular atrophy: the Australian experience. Genet Med. 2020;22(3):557-65.