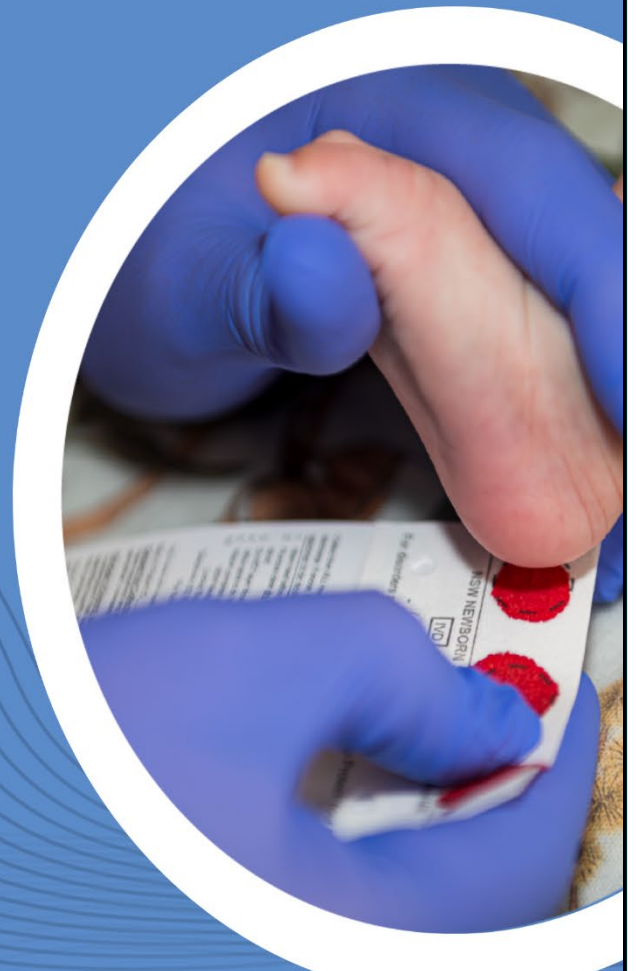


2025

Summary of Recommendations

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



Section 1: Screening for *SMN1* as part of (newborn) screening in SMA

Recommendation 1.1

Evidence based recommendation

We recommend that newborn screening for SMA should be performed on the routine newborn dried blood spot with absence of exon 7 on *SMN1* as the target analyte.

Grade of recommendation Strong, for

Recommendation 1.2

Consensus recommendation

The screening method selected by the screening program should have a sensitivity of $\geq 95\%$ for the detection of *SMN1* exon 7 absence (0 *SMN1* copies) using suitably validated quantitative and qualitative assays

Recommendation 1.3.

Consensus recommendation

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).

Section 2: Screening for *SMN2* copy number as part of (newborn) screening in SMA

Recommendation 2.1.

Consensus recommendation

SMN2 copy number should be performed expeditiously, ideally as part of newborn screening processes using suitably validated quantitative assays but the result should not delay notification of the absence of exon 7 on *SMN1*.

Recommendation 2.2.

Consensus recommendation

Newborn screening programs should establish a clinical referral pathway that includes simultaneous notification of a screen positive result to a paediatric neurology specialist and a local healthcare practitioner.

Section 3: Confirming a diagnosis of SMA in screen positive newborns

Recommendation 3.1

Evidence based recommendation

Diagnostic testing should include confirmation of an absence of exon 7 on *SMN1* (i.e. zero copies of *SMN1*).

Grade of Recommendation: Strong, for

Recommendation 3.2

Consensus recommendation

Diagnostic testing using suitably validated assays, from whole blood samples or repeat dried blood spot from a recalled infant should include *SMN2* copy number as a guide to prediction of clinical severity and to facilitate therapeutic decision making.

Recommendation 3.3

Consensus recommendation

Diagnostic results for *SMN1* should be available as quickly as possible, and at maximum of 7 days of receipt of the sample by the diagnostic laboratory.

Recommendation 3.4

Consensus recommendation

A diagnosis of SMA (including *SMN1* and *SMN2* copy number results) should be available to clinical services as quickly as possible. This should be completed within 30 days of birth to enable timely treatment.

Section 4: Managing uncertain, false positive and false negative screening results

Recommendation 4.1

Consensus recommendation

For newborns with a false positive, false negative **or** uncertain screening result, a case review with communication and collaboration between screening, diagnostic and clinical services should be conducted to understand the aetiology of results and explained to families.

Recommendation 4.2

Consensus recommendation

If there is a difference in *SMN1* and/or *SMN2* copy number results between screening and diagnostic assays, retesting for *SMN1* and/or *SMN2* copy number with another method/laboratory should be considered.

Recommendation 4.3

Consensus recommendation

If there is uncertainty as to the diagnosis of SMA the child should be clinically followed up by a paediatric neurologist until diagnostic certainty is reached.

Section 5: Communicating a SMA screen positive result to families

Recommendation 5.1

Consensus recommendation

Screen positive result should be disclosed to the family within ≤ 2 working days (of notification to healthcare services).

Recommendation 5.2

Consensus recommendation

Screen positive newborns should be offered a clinical review within paediatric neurology/neuromuscular services within ≤ 2 working days, from the time of screen positive disclosure.

Recommendation 5.3

Consensus recommendation

Culturally safe care is required by healthcare practitioners when disclosing screening results to families from Aboriginal, Torres Strait Islander, Pacific Islander, Māori or other culturally and linguistically diverse backgrounds. If the healthcare practitioner is not bilingual, a professional interpreter should be used and advice and support sought from Indigenous Health Liaison professionals (which may include a First Nations nurse, midwife or healthcare practitioner) where relevant and appropriate.

Section 6: Assessments required at diagnostic evaluation of the newborn

Recommendation 6.1

Consensus recommendation

The following assessments should be completed immediately as part of the diagnostic and clinical evaluation of the newborn, who screens positive for SMA.

- Neurological examination.
- Venous sampling for quantification of *SMN1* exon 7 on whole blood.
- Venous sampling for determination of *SMN2* copy number on whole blood OR repeat dried blood spot for confirmation of *SMN2* copy number.

Section 7: Provision of information and support for families after confirming the diagnosis of SMA in the (screen positive) newborn

Recommendation 7.1

Consensus recommendation

The process of disclosing a diagnosis of SMA to families should occur with a paediatric neurologist when *SMN1* (diagnostic) confirmation is received, regardless of the availability of *SMN2* copy number result.

Recommendation 7.2

Consensus recommendation

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 reflect the contemporary care landscape and are nationally consistent.

Recommendation 7.3

Consensus recommendation

Culturally safe care is required by healthcare practitioners when disclosing diagnostic results to families from Aboriginal, Torres Strait Islander, Pacific Islander, Māori or other culturally and linguistically diverse backgrounds. If the healthcare practitioner is not bilingual, a professional interpreter should be used and advice and support sought from Indigenous Health Liaison professionals (which may include a First Nations nurse, midwife or healthcare practitioner) where relevant and appropriate.

Section 8: Immediate post diagnostic care for newborns and infants receiving a diagnosis of SMA through a newborn screening program

Recommendation 8.1

Consensus recommendation

For screen positive newborns who demonstrate signs and symptoms of SMA (consistent with disease onset i.e. clinically manifest), a paediatric neurologist should discuss options for immediate treatment with SMN augmenting treatments, with the family.

Recommendation 8.2.

Consensus recommendation

For newborns with diagnostic confirmation of SMA and 1, 2 or 3 *SMN2* copies and who are presymptomatic (i.e. clinically silent), a paediatric neurologist should discuss options for immediate SMN augmenting treatments, with the family.

Recommendation 8.3

Consensus recommendation

In the absence of comparative data, single agent treatment i.e. monotherapy at initiation of therapeutic intervention is recommended, started within paediatric neurology treatment centre.

Recommendation 8.4.

Consensus recommendation

Newborns with diagnostic confirmation of SMA who are unable to access approved and reimbursed treatments or chose not to be treated immediately, should have clinical follow-up

with a minimum of 3 monthly assessments for the first two years from diagnosis, and minimum 6-monthly thereafter.

Recommendation 8.5.

Consensus recommendation

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 clinical genetics services).