National Policy ☐ National Procedure ☐ National Protocol ☐ National Guideline ☐
National Clinical Guideline ☒

Management of Infants with a Suspected Diagnosis of Spinal Muscular Atrophy (SMA) through the National Newborn Bloodspot Screening Programme (NNBSP)

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Management of Infants with a Suspected Diagnosis of Spinal Muscular Atrophy (SMA) through the National Newborn Bloodspot Screening Programme (NNBSP)
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Short summary:
The management of infants with a suspected diagnosis of Spinal Muscular Atrophy (SMA) Through the National Newborn Bloodspot Screening Programme (NNBSP)
Description:
To provide an evidence-based, nationally standardised, time-critical clinical pathway for the management of infants who screen positive for a suspected diagnosis of Spinal Muscular Atrophy (SMA) through the Irish National Newborn Bloodspot Screening Programme (NNBSP).

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Contents

1.0 Guideline Development Group members:

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- Consultant Paediatrician with interest in Neurodevelopment: Prof. Denise McDonald
- Consultant Clinical Geneticist:
- Specialist Neuromuscular Nurse: Ms Hannah McLoughlin
- Gene Therapy Coordinator: Ms Derval Reidy
- Physiotherapist: Ms Maeve O'Hanlon
- Parent and Advocacy Representatives (SMA Ireland): Ms Bronagh Noonan

2.0 Aim:

Management of infants with a suspected diagnosis of Spinal Muscular Atrophy (SMA) Through the National Newborn Bloodspot Screening Programme (NNBSP)

- To provide an evidence-based, nationally standardised, time-critical clinical pathway for the management of infants who screen positive for a suspected diagnosis of Spinal Muscular Atrophy (SMA) through the Irish National Newborn Bloodspot Screening Programme (NNBSP), ensuring/ supporting:
  - Timely initiation of bridging therapy with Risdiplam as soon as possible following screen-positive notification pending MLPA result and availability of Onasemnogene Abeparvovec (Zolgensma), if indicated.
  - Rapid confirmatory testing of SMN1 deletion for infants with a suspected diagnosis of SMA, through multiplex ligation-dependent probe amplification (MLPA)
  - Rapid quantification of SMN2 copy number, using MLPA, to inform prognosis and treatment planning.

- Timely (as soon as possible) administration of Onasemnogene Abeparvovec (Zolgensma) for confirmed cases of SMA, in the Gene Therapy Suite, St. James's Hospital, or alternatively continuation of Risdiplam where appropriate.
- Appropriate clinical follow up and multidisciplinary care.
- Standardised roles, monitoring, safety procedures, and communication pathways.

3.0 Target Users:

- All healthcare professionals who are involved in the diagnosis and care of infants with a suspected diagnosis of SMA, including Paediatricians, Neurologists, Neonatologists, Clinical Geneticists, Nursing staff, Allied health professionals, Gene therapy teams and the National Newborn Bloodspot Screening Programme.

4.0 Target patient Population:

Infants identified as screen-positive for SMA through the National Newborn Bloodspot Screening Programme (NNBSP).

- Includes pre-symptomatic and minimally symptomatic infants.

5.0 Associated documents:

This guideline should be read and used in conjunction with the following document(s):

- HSE Managed Access Protocols for Onasemnogene Abeparvovec, Risdiplam and Nusinersen
- National Newborn Bloodspot Screening Programme Screening Pathway for Spinal Muscular Atrophy (SMA) and clinical pathway for babies who screen positive for SMA

6.0 Definitions / Abbreviations:

Spinal Muscular Atrophy (SMA): A severe autosomal recessive neuromuscular disorder caused by biallelic loss or dysfunction of the Survival Motor Neuron 1 (SMN1) gene, leading to progressive degeneration of alpha motor neurons in the anterior horn of the spinal cord.

SMN1 Gene: The primary gene responsible for producing full-length SMN protein. Homozygous deletion or pathogenic variants in SMN1 cause SMA.

SMN2 Gene: A paralog of SMN1 that produces a reduced amount of functional SMN protein due to alternative splicing. The number of SMN2 gene copies (2, 3, 4+) partially modifies SMA disease severity, with higher copy numbers associated with milder phenotypes.

MLPA (Multiplex Ligation-dependent Probe Amplification): A molecular diagnostic technique used to confirm: SMN1 deletion status; SMN2 copy number.

National Newborn Bloodspot Screening Programme (NNBSP): A national population-based screening programme using dried blood spot samples to identify presymptomatic infants with serious congenital conditions, including SMA.

Bridging Therapy: Interim disease-modifying treatment initiated before confirmatory diagnostic testing for SMA is complete and while treatment planning is ongoing, to prevent irreversible motor neuron loss while awaiting MLPA results and while waiting to obtain Onasemnogene Abeparvovec (Zolgensma), if applicable. In this pathway, Risdiplam is the bridging therapy.

Risdiplam: An orally administered SMN2 splicing modifier that increases full-length SMN protein production. Used as: Long-term therapy for SMA types 1–3; Immediate bridging therapy in SMA newborn-screening pathways.

Onasemnogene Abeparvovec (Zolgensma): A single-dose gene replacement therapy using an adeno-associated virus serotype 9 (AAV9) vector to deliver a functional copy of SMN1 to motor neurons. Given as a one-time intravenous infusion in a specialised gene therapy suite.

Gene Therapy Suite: A dedicated facility within St. James's Hospital, Dublin, equipped for aseptic preparation, administration, and monitoring during AAV-based gene therapy infusion.

Prednisolone Pre-Treatment: A corticosteroid regimen initiated three days before Zolgensma to mitigate immune-mediated liver inflammation associated with AAV9 gene therapy vectors.
Pre-Symptomatic Treatment: Therapy provided before the onset of clinical symptoms, shown to improve motor milestones, ventilation-free survival, and developmental outcomes in SMA.
CHOP INTEND (Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders): A validated motor scale to assess neuromuscular function in infants with SMA, used for baseline and follow-up assessments.
HINE-2 (Hammersmith Infant Neurological Examination – Part 2): A structured neurological assessment tool measuring motor milestones in infants, widely used in SMA treatment monitoring.
AAV9 (Adeno-Associated Virus Serotype 9): The viral vector used in Zolgensma to deliver the functional SMN1 gene. AAV9 has tropism for motor neurons and crosses the blood–brain barrier.
ALT / AST: Liver enzymes (alanine aminotransferase (ALT), aspartate aminotransferase (AST)) measured during monitoring for potential hepatotoxicity post-Zolgensma.
Troponin-I: Cardiac biomarker used to detect rare AAV9-mediated myocarditis or myocardial stress following gene therapy.

7.0 Guidance

SMA is an autosomal recessive neuromuscular disorder resulting from loss of SMN1 function. Until recently, progressive motor neuron degeneration led to early mortality. Access to gene therapy, antisense oligonucleotides, and SMN2-modifying agents has transformed outcomes.

Detection of SMA by newborn bloodspot screening uses quantitative polymerase chain reaction (qPCR)-based methods to detect homozygous deletions of the SMN1 gene. Samples which return a screen positive result for SMN1 deletion undergo confirmatory testing using multiplex ligation-dependent probe amplification (MLPA) to confirm SMN1 deletions and to quantify SMN2 copy number to inform prognosis and treatment planning. Multiplex ligation-dependent probe amplification (MLPA) is regarded as the gold standard for diagnosis of SMA.

Newborn screening enables treatment before symptom onset, improving survival and motor development.

The aim of newborn screening for SMA in Ireland is to detect babies with 5q SMA caused by a homozygous deletion of exon 7 of the SMN1 gene (accounting for approximately 95% of SMA cases). Newborn screening cannot currently detect cases of SMA caused by compound heterozygous variants of SMN1 or cases of non-5q SMA. Additionally, as with all screening programmes, there is the potential for false negative screening results. As such, a negative screening result for SMA does not preclude the diagnosis.

8.0 Recommendations

1. For all babies who screen positive through PCR-based screening via the NNBSP, start Risdiplam bridging therapy as soon as possible, ideally same day of positive newborn screen result.
2. Send blood sample for confirmatory MLPA testing (SMN1 deletion status) and quantification of SMN2 copy number to inform prognosis and treatment planning.
3. Blood sample for MLPA should be sent, ideally on the same day as screen notification, or within 1 working day, where possible.
4. For all infants where MLPA confirms a diagnosis of SMA, begin Zolgensma eligibility procedures.
5. If MLPA result is negative/ normal, arrange repeat MLPA without delay and continue bridging therapy pending MLPA result.
6. If repeat MLPA result remains negative/ normal, screen is considered a false positive and the parents can be counselled and reassured and the baby discharged from the screening programme.

7. For infants with confirmed diagnosis of SMA, if appropriate, treat with Zolgensma by approximately Day 30 of life.
8. Initiate prednisolone 1 mg/kg/day three days prior to Zolgensma infusion. Risdiplam is discontinued after Zolgensma infusion.
9. Implement weekly post-infusion monitoring of liver function, troponin I, and FBC.

9.0 Conditional Recommendations

1. Provide early input from physiotherapy, dietetics, speech & language therapy and medical social worker. Clinical psychology support and genetic counselling are recommended following MLPA confirmation.
2. Advise strict infection avoidance before gene therapy.

10.0 Clinical Pathway

Day 0: Positive SMA Screen notified to Paediatric Neurology Team by National Newborn Bloodspot Screening Laboratory (NNBSL) → Liaise with parents / inform local neonatal or paediatric team / next working day Neurology Review → Start Risdiplam bridging therapy → Send MLPA

⬇

Days 1–7: Continue bridging therapy, baseline monitoring, parental counselling

⬇

Day 7: MLPA results received (SMN1 deletion + SMN2 copies)

⬇

SMA confirmed → Begin Zolgensma eligibility procedures and continue bridging therapy

⬇

Day 21: Start prednisolone for pre-treatment

⬇

Day 24-30: Zolgensma infusion (St. James's Gene Therapy Suite)

⬇

Post-Treatment: Discontinue bridging therapy post Zolgensma infusion. Monitor infant with weekly LFTs/troponin/CBC + developmental follow-up.

11.0 Detailed Pathway Steps

Screen-Positive Notification

Positive SMA Screen notified to Paediatric Neurology Team by National Newborn Bloodspot Screening Laboratory (NNBSL)

12.0 Actions Required

Paediatric Neurology Team to immediately notify:

- Paediatric Neuromuscular Lead (CHI)
- Local consultant paediatrician
- Neuromuscular nurse specialist
- Gene therapy coordinator

Communication with parents(s)

Paediatric Neurology Team to contact the parents without delay (same day as initial notification of screen positive result, but not on Friday if infant cannot be reviewed same day/ next day) and inform them of the screening result, implications of same, next steps, and provide parent with information leaflet.

Parent Counselling

Information points to include:

- Reason for suspected diagnosis of SMA (positive NBS result).
- Requirement for MLPA and expected turnaround time.
- Risdiplam as bridging therapy.
- Expected timelines.
- Treatment options (if SMA confirmed on MLPA).
- Anticipated improvement rates with early treatment (if SMA confirmed on MLPA).

Clinical Review - CHI

- Same day/ next feasible working day review.
- Medical Social Worker review if available.
- Assess for tone, reflexes, swallowing, and respiratory function. If indicated, arrange for swallow assessment and respiratory referral.

Bridging Therapy

- Start Risdiplam immediately. Inform CHI pharmacy to order a single bottle of Risdiplam directly from Roche. It will require 24 hours to dispatch to CHI pharmacy. Risdiplam can be dispensed directly from CHI hospital pharmacy.
- Weight-based dosing as per product information.
- Document time of first administration.

Confirmatory Testing

- Draw blood for MLPA (SMN1 + SMN2).
- Turnaround time: 5-7 days.

Baseline Investigations

To avoid delays before Zolgensma:

- LFTs
- FBC
- Troponin-I
- AAV9 serology
- Coagulation screen

13.0 Days 1–7: Pre-Confirmation Management

- **Continue Risdiplam**
- Monitor feeding/tolerability.

Multidisciplinary Care - CHI

- Physiotherapy: baseline motor function
- Dietetics: nutrition optimisation
- Speech and Language therapy (SALT): feeding review if needed
- Medical Social work: early support

14.0 Day 7 – Confirmatory MLPA Results

Interpretation

- SMN1 homozygous deletion confirmed = Result interpreted in context of clinical status to help determine SMA diagnosis (presymptomatic or symptomatic)
- SMN2 copy number = Result combined with clinical status will help determine SMA phenotype and inform treatment choice.

Note: If MLPA results is normal/ does not confirm SMN1 homozygous deletion, paediatric neuromuscular team to communicate result to parents, arrange repeat MLPA without delay and continue bridging therapy. If repeat MLPA is normal, parents can be counselled, bridging therapy can be discontinued and the baby can be discharged from the Newborn Bloodspot Screening Programme.

Communication – SMA diagnosis confirmed

- Consultant neurologist or neuromuscular nurse communicates the result to the parents same day, and arranges next steps and contacts local paediatrician if infant born in RPU or LPU.
- Document confirmatory diagnosis
- Update treatment plan and timeline

Days 7–24: Pre-Zolgensma Preparation

Eligibility Screening

- Infant must be clinically stable and infection-free
- Repeat labs (LFTs, CBC, troponin)
- Anti-AAV 9 serology result

Treatment application

- Application for Zolgensma treatment through MMP pathway (if AAV9 negative and 3 or less SMN2 copies)
- Application for Risdiplam treatment continuation through MMP pathway, if clinically appropriate or if AAV9 positive or 4 SMN2 copies.

Scheduling

- Gene Therapy Coordinator books:

  - Infusion date
  - Gene Therapy Suite
  - Required staff
  - Pharmacy preparation

Infection Prevention and Control pre Zolgensma Treatment

- No visitors except immediate household
- Avoid crèche, public areas, large gatherings

Day 24-30 – Zolgensma Infusion

Location

- Patient admitted to CHI
- Zolgensma infusion in Gene Therapy Suite, St. James's Hospital

Medication

- Start prednisolone 1 mg/kg/day on day prior to treatment
- Continue through infusion and for minimum 30 days post-infusion

Procedure

- Single-dose IV infusion over 60 minutes
- Continuous observation for ≥4 hours
- To remain in Gene Therapy Suite in St James's Hospital for one hour post administration
- Monitor for vomiting, irritability, pyrexia

- Inpatient observation in CHI for 7-10 days post infusion and as clinically appropriate. Isolation room required to control viral vector shedding.
- Discontinue Risdiplam after completion of Zolgensma infusion.

15.0 Discharge

- Steroid continuation
- Weekly blood monitoring (4-8 weeks)
- Emergency contact guidance

16.0 Zolgensma Post-Treatment Monitoring (as per SMPC)

Weekly Monitoring for 4–8 Weeks, to be arranged in either CHI or locally as appropriate. Local Paediatric service to contact neuromuscular CNS with results.

- LFTs
- FBC
- Troponin I
- Consider viral PCRs if intercurrent illness

Physiotherapy & Developmental Review in CHI

- HINE-2 or CHOP INTEND scoring
- Monthly early motor reviews initially

Vaccinations

- Defer live vaccines until steroids discontinued
- Routine vaccines may proceed once medically cleared

17.0 Safety, Risk Mitigation and Escalation

Hepatotoxicity (Common)

- If ALT/AST rise x 3 ULN:
    - Increase prednisolone dose
    - Delay steroid weaning
    - Repeat labs 48–72 hours

Cardiac Monitoring

- Troponin elevations → cardiology referral

Thrombocytopenia

- Re-check FBC weekly
- Severe thrombocytopenia (<50x109/L): haematology consult

Family follow-up

- Parental SMN1 carrier status confirmation
- Genetic counselling referral
- Clinical psychology referral

18.0 Implementation Plan

- National training webinars for clinicians
- Standardised Risdiplam prescribing protocols
- Checklist for Zolgensma eligibility
- Integration with new MMP application process
- HSE communication strategy for families and clinicians

19.0 Supporting Documents:

Health technology assessment of the addition of spinal muscular atrophy (SMA) to the National Newborn Bloodspot Screening Programme Status: Published on 27 Nov 2023

20.0 References:

Early Identification & Newborn Screening

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2. Glascock J, Sampson J, Maldonado M, et al. SMA Newborn Screening and Early Treatment Recommendations. J Neuromuscul Dis. 2020;7(2):97–109.
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2. Prior TW. Carrier screening for spinal muscular atrophy. Genet Med. 2008;10(11):840–842.

Risdiplam (Bridging Therapy)

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3. Day JW, Chiriboga CA, Crawford TO, et al. Early treatment of presymptomatic SMA with onasemnogene abeparvovec. Nat Med. 2021;27:229–232.
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Timing of Treatment (Pre-symptomatic)

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3. Dangouloff T, Servais L. Newborn screening programs for SMA: why early is better. Neuromuscul Disord. 2019;29:1031–1038.

Monitoring, Steroid Use, & Safety

1. Chand DH, Zaidman C, Arya K, et al. AAV9 gene therapy clinical monitoring for hepatotoxicity and safety. Mol Ther. 2021;29:1415–1421.
2. Mendell JR, Al-Zaidy S. Post-gene therapy liver monitoring in SMA. J Pediatr. 2020;221:223–228.
3. Lowes LP, Alfano LN, Arnold WD, et al. Post-onasemnogene abeparvovec monitoring guidelines. Neurol Genet. 2021;7(5):e615.

Motor Scales (CHOP-INTEND, HINE-2)

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3. Kolb SJ, Coffey CS, Yankey JW, et al. Natural history of infantile SMA with and without treatment. J Child Neurol. 2017.

Health Service Delivery, Implementation & Pathways

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Ethical, Logistical, and Operational Considerations

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2. Darras BT. SMA treatment—new standards of care with gene therapy. Pediatr Neurol. 2021; 118:3–4.

21.0 Roles and Responsibilities

It is the responsibility of:

National Newborn Bloodspot Screening Programme:

- Deliver and quality assure newborn bloodspot screening for SMA.
- Ensure immediate notification of paediatric neurology when an infant screens positive for SMA.
- Ensure timely delivery of confirmatory testing via MLPA.

Lead Consultant Paediatric Neurologist (CHI):

- For infants with positive NBS result for SMA, communicate screen result and next steps to parent and local paediatrician on call
- Provide/ offer same day/ next working day clinical review for screen positive infants.
- Initiate Risdiplam and request MLPA for infants with suspected SMA.
- Communicate MLPA result to parent.
- In screen detected babies, if MLPA result is normal/ negative, arrange repeat MLPA and repeat dried bloodspot sample.
- If SMA confirmed, communicate diagnosis to parents, determine treatment plan, request Zolgensma.
- Oversee prednisolone protocol and post-treatment monitoring.
- Initiate familial testing as appropriate (e.g. carrier testing in parents)

Department of Clinical Genetics (DCG) labaratory CHI:

- Perform MLPA testing.
- Notify relevant teams immediately when testing has been performed.
- Issue MLPA report.
- Maintain QA standards.

Clinical Geneticist and Genetic counselling:

- Provide genetic counselling and recurrence-risk information.
- Initiate familial testing as appropriate (e.g. sibling testing)
- Document results and liaise with clinical teams.

Neuromuscular Nurse Specialist / SMA Coordinator:

- Primary family contact.
- Provide education on bridging therapy, steroids, and monitoring.
- Coordinate appointments and pathway logistics.
- Support documentation and follow-up.

Gene Therapy Coordinator (St. James's Hospital):

- Schedule Zolgensma infusion.
- Ensure all pre-infusion labs and requirements are completed.
- Coordinate pharmacy, nursing, and suite availability.
- Maintain infusion records and pathway timelines.

Gene Therapy Team (St. James's):

- Prepare and administer Zolgensma.
- Provide continuous observation and manage immediate adverse events.
- Deliver discharge guidance and post-infusion instructions.

Pharmacy (CHI and St. James's):

- Verify and dispense Risdiplam.
- Prepare Zolgensma under aseptic conditions.
- Maintain cold chain and regulatory compliance.

Allied Health Professionals (CHI):

- Physiotherapy: Baseline motor assessment and early intervention.
- Dietetics: Optimise nutrition pre- and post-infusion.
- SALT: Review feeding/swallowing.
- Social Work: Provide practical and emotional support.

- Clinical psychology: Provide practical and emotional support.

Parents / Guardians:

- Administer Risdiplam correctly.
- Follow infection-prevention guidance.
- Attend scheduled appointments and report concerns.

Primary Care:

- Provide general health monitoring.
- Avoid live vaccines until cleared.
- Refer urgently if concerns arise.

HSE / National Oversight:

- Provide governance and monitoring of clinical treatment pathway performance.
- Support audit, quality assurance, and updates to the pathway/ guideline.

22.0 Communication and Training

Communication and Dissemination:

This guideline will be published on the National Clinical Respository. Information about its publication will be circulated to relevant departments.

- Where appropriate, the document owner will advise healthcare professionals in Ireland but outside CHI by email of the publication.

11.2 Training:

Indicate if a specific training session is required or if included as a named document in the unit/service-based induction

23.0 Monitoring & Evaluation

- The impact of this guideline on patient care and outcomes will be monitored by noting trends in clinical practice, issues identified through safety huddles, incident and near miss reporting, patient/parent complaints, and where relevant, specific KPIs:
  - For screen detected infants, time from screen-positive result to first dose of Risdiplam (target: same day).
  - Turnaround time of MLPA (target: ≤7 days).
  - Time from birth to Zolgensma infusion (target: day 24-30).
  - Incidence of elevated transaminases requiring steroid escalation.
  - Motor outcome scores at 6 and 12 months.
  - Parent satisfaction and support pathway metrics.
- Department managers are responsible for ensuring their teams are aware of and practising in accordance with the guideline and will relay feedback on the guideline to the owner/author.
- CHI staff are responsible for practising in accordance with the guideline, and for escalating feedback or concerns about the guideline to their department manager.
- Evaluation of the guideline and any change requests to the content will be made in response to emerging evidence, trends in practice, incidents or complaints and/or user feedback.
- The NNBSP Clinical and Quality Assurance (CQA) Group will carry out early and periodic evaluations of the performance and outcomes of the SMA screening programme. CHI will engage with the NNBSP to support delivery of this evaluation.

24.0 Guideline Review Process

National Clinical Lead for Children, NCPPN	February	2026
Paediatric Clinical Advisory Group	January	2026
National Clinical Advisor & Group Lead Children and young people	February	2026