

BPNA Statement on the Urgent Need for SMA Newborn Screening in the UK

The British Paediatric Neurology Association (BPNA) with the BPNA Muscle Interest Group (MIG) is committed to a world where every child with a neurological condition receives the care and support they need to reach their full potential. Central to this mission is our advocacy for universal and early access to disease-modifying treatments whenever they are available and Spinal Muscular Atrophy (SMA) is a condition where these treatments are extremely effective.

We believe every child born with SMA deserves the maximum benefit afforded by early intervention as unequivocally demonstrated by every single study published so far, using any one of the 3 approved disease modifying therapies. Currently, this is not possible in most of the UK because a universal newborn screening (NBS) programme has not yet been established.

Every year around 70 babies are born with SMA in the UK and NHS has made substantial investment in disease-modifying therapies, improving survival; however, without newborn screening this survival is often accompanied by very significant avoidable morbidity and long term-disability, attributable to the diagnostic delay which is commonly encountered in clinical practice. This has substantial long-term impact not only on the child but also their wider family, and the NHS resources. Evidence from other newborn screening programmes consistently shows that optimal outcomes, including the possibility of normal or near normal development which can only be achieved with presymptomatic treatment. When these costly therapies are initiated late, the outcomes are suboptimal for children, resulting in significantly poor value for NHS investment.

Furthermore, delayed diagnosis places an unavoidable burden on clinicians who must manage preventable disease progression, with the knowledge that earlier detection would have altered outcomes.

A Critical Delay in Implementation

The UK National Screening Committee (UK NSC) began its review into SMA screening in November 2022. More than three years later, we have significant concerns regarding the lack of progress. The human cost of this delay has been starkly highlighted by the recent, heartbreaking news shared by Jesy Nelson regarding her daughters' diagnosis, a situation that underscores the vulnerability of many children and families trapped by current diagnostic timelines.

While the UK NSC announced an In-Service Evaluation (ISE) in August 2023 to assess feasibility, progress remains unacceptably slow:

- **Delayed Start:** The first baby is not expected to be screened in England until 2027 at the earliest.
- **Partial Coverage and inequity:** The ISE will use a "stepwise" approach, and, will cover only two third of newborns in England. In addition, it does not include Wales and Northern Ireland, creating inequity in accessing life-changing early diagnosis conflicting with NHS commitments to equity and fairness.

Precedent for Change

A clear path forward exists. **Scotland has set a precedent for timely action**, announcing a national programme in September 2025 to gather evidence on SMA screening effectiveness. Remarkably, Scotland has moved from announcement to implementation in just six months, with **the first baby due to be screened in March 2026**.

In contrast, the wider UK approach is becoming increasingly isolated. Robust clinical and economic evidence supports SMA screening; **nearly two-thirds of Europe already screen for the condition, and 100% of newborns are screened in the United States, Australia and Canada.**

Our Call to Action

Based on current trajectories, formal UK-wide screening is unlikely to be delivered before the end of the decade. This represents a failure to parents and a contradiction to one of the 3 major shifts outlined in the NHS 10-year plan published in 2025: sickness to prevention, as well as to the NHS long term plan emphasising the use of new medicines to deliver better care and value of taxpayers. Without an urgent change in approach, hundreds more newborns will face delayed SMA diagnoses, leading to irreversible motor neuron loss, long-term disability and reduced life expectancy.

The BPNA with the BPNA MIG calls for:

1. **Accountability:** The UK Newborn Screening Committee must be held to account for these protracted timelines.
2. **Immediate Implementation:** The In-Service Evaluation (ISE) must be accelerated and expanded to ensure as many babies as possible are screened immediately.
3. **Evidence-Based Action:** Given the wealth of international evidence, there is a very strong case for bypassing the complexities of the current ISE and immediate implementation of screening nationwide without further delay.

The absence of SMA newborn screening denies families the opportunity for access to therapies that can fundamentally rewrite a child's future. We must act now.