Every Moment Matters
The case for introducing newborn screening for spinal muscular atrophy key recommendations

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What is SMA and who's affected?

Spinal Muscular Atrophy (SMA) is a rare but debilitating genetic disease that has a devastating impact on those affected and their families.[1]

For newborns diagnosed with SMA Type 1, the most common form of the condition accounting for **60% of cases**, many experience significant and irreversible disability because of delayed diagnoses and access to treatment.[2,3]

SMA is the leading genetic cause of infant death in the UK, and **affects approximately 68 babies every year**[4]. Without timely treatment, more than 90% of babies with SMA Type 1 will die or need permanent ventilation by their second birthday, while most people with SMA Type 2 will never walk independently.[2,6]

Diagnosing and treating SMA at the earliest possible point provides the strongest opportunity to prevent disease progression.

68 babies are estimated to be born with SMA in the UK every year.[5]

Between **1 in every 40 to 60** are carriers for the defective gene that leads to SMA in people. This means that incidence of SMA in the UK is estimated to be 1 in 10,000.[7,8]

It is estimated that there are **1,350 people across the UK living with SMA**.[5]

The case for Newborn Screening

Newborn screening for SMA is the fastest route to early and pre-symptomatic diagnosis, however, it is not currently included within the NHS Newborn Screening Programme. Under current pathways, unless there is a known familial association of SMA, diagnoses are made after children start to develop symptoms, which is typically within the first six months from birth.[9]

For babies born with SMA Type 1, the average age of diagnosis is **6.3 months, by which time 95% of motor neurons can be lost.**[9]

In the absence of newborn screening and even when treatment is initiated following diagnosis after symptom onset, most infants with SMA Type 1 will:

Require mechanical ventilation

Need nutritional support

Be given round the clock care

However, early diagnosis and pre-symptomatic treatment - made possible through newborn screening - provides a vital opportunity to prevent avoidable disability and mortality among newborns with SMA.[10]

> **Screening for SMA at birth will significantly increase a newborn's chance of survival. Today, in the UK, we are not screening our children for SMA even though we know how it works and we know how to use it. This must change.**
>
> **Professor Laurent Servais:** Professor of Paediatric Neuromuscular Diseases at the MDUK Oxford Neuromuscular Centre, and Chair of the UK SMA Screening Alliance[9]

> **At SMA UK we believe not having SMA included on the newborn screening programme is unethical, as children are living with exceptional and complex needs that can be minimised or even prevented. SMA UK are working to achieve the earliest possible introduction of newborn screening for SMA in the UK. Through this simple mechanism the NHS will deliver the best outcomes from treatment and reduce future healthcare costs over the lifetime of that person.**
>
> **Giles Lomax:** Chief Executive, SMA UK

The burden of SMA on patients and carers

In addition to the immediate physical impacts of SMA, the condition also generates considerable, multifaceted emotional and economic costs, many of which are avoidable.

Almost **one third of people** with SMA see **more than 10 HCPs per year.**[11]

On average, **three unpaid caregivers** provide support to someone with SMA Type 1 and Type 2.[11]

For those caring for a child with SMA Type 1, **74% report that one or more caregivers had given up work** completely.[11]

A study of 86 children with SMA – 26.7% having SMA Type 1 and 73.3% having SMA Type 2 and 3 – found that the **annual average cost was €54,295 per child** in the UK.[12]

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The UK is behind the curve

The UK is an outlier in relation to newborn SMA screening, which is currently in place within the United States, more than half of European Union member states, and nearly two-thirds of Europe geographically, including countries such as Russia, Turkey and Ukraine.[1,2]

Map showing the European countries that have nationally recommended and implemented newborn screening programmes for SMA, and where pilot programmes have covered 100% of the population.

The impact of delaying newborn screening for SMA

Treatments for SMA were made routinely available on the NHS in June 2019. Since then, the UK SMA Newborn Screening Alliance's Monthly Barometer estimates that, as of January 2024, **332 newborn babies and their families have been impacted by avoidable and irreversible damage** due to SMA, which could have been avoided by the introduction of newborn screening.

Recommendations and calls to action for the UK NSC and UK government

The UK NSC should announce details of its in-service evaluation (ISE) of newborn screening for SMA. To avoid the deepening of inequalities in access to newborn screening witnessed across **the UK the ISE should cover all four UK-nations or be implemented at a pace that would enable any inequities in screening to be short-lived.**

The UK Government must ensure **sufficient funding is available – to enable effective, urgent and UK-wide implementation and delivery of newborn screening for SMA** – both in the ISE phase and following an anticipated future positive full screening recommendation.

Ministers responsible for newborn screening across all four nations of the UK and the UK National Screening Committee should work with patients, families, clinicians and the patient group community to **agree on clear and regular milestones for its review of newborn screening for SMA,** to ensure its timely introduction.

The UK NSC should set out opportunities for collaboration with the SMA and rare disease community on the practical implementation of newborn screening – both in the ISE phase and following a future positive recommendation – **to ensure system readiness for the adoption and equitable implementation of screening across all four nations.**

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