Every Moment Matters:
Implementing newborn screening for Spinal Muscular Atrophy in the UK

November 2023

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Contents

Foreword

Spinal muscular atrophy (SMA) is an extremely rare but debilitating genetic condition that affects at least one newborn baby every five days in the UK. Children born with the most common form of the condition (Type 1) face life limiting disability and disease progression. If left undiagnosed and untreated, many children with SMA do not live beyond their second birthday.

As a condition that has a profound impact on those affected and their families, it is imperative that we use the tools we have at our disposal to prevent avoidable disability and mortality. Fortunately, treatment options exist that can prevent disease progression and enable children born with SMA to lead healthy and independent lives if initiated before symptom onset. Without doubt, newborn screening offers the best chance we have for achieving early intervention and transforming the life that somebody affected by SMA will lead.

Without screening however, a diagnosis can only be made once babies start to display symptoms which usually appear after 6 months, unless there is a known family history of the condition which is usually not the case. However, by this time, babies will suffer significant loss of motor neurons which cannot be regenerated, and therefore the disability experienced cannot be reversed. This makes the UK a significant outlier in comparison to other European countries – with SMA currently being screened for within more than half of European Union member states, and nearly two-thirds of Europe geographically. It is therefore urgent that we challenge the status quo in the UK and rectify this unacceptable inequality that newborns face if they are diagnosed with SMA in the UK.

In light of this we are pleased to see that the UK National Screening Committee is conducting a review to decide whether to include SMA within the NHS newborn bloodspot programme. If included, babies will be screened at birth and can be offered treatment and support before they experience symptoms. This is a huge opportunity to drastically improve the lives of these children and their families, and we hope to see screening recommended and implemented imminently.

More broadly, newborn screening represents a vital tool in meeting UK-wide rare disease policy ambitions, with all four respective national Action Plans emphasising the importance of screening initiatives in supporting earlier detection and addressing the diagnostic odyssey experienced by so many with a rare disease. SMA represents a clear and tangible example of how newborn screening can transform the outcomes of people living with one of these rare conditions, and an opportunity to set out a blueprint for how the processes for adding conditions to the NHS bloodspot programme can be streamlined to deliver both robust and timely decisions.

We therefore support the Every Moment Matters campaign which seeks to ensure screening is introduced at pace and without delay – in light of the wealth of available evidence to support its introduction - to bring the UK in line with so many other countries that already screen for this condition, and to ensure every part of the system is aligned to allow those affected with access to interventions that offer the best chance of a healthy life.

We hope that the personal stories and recommendations from the contributors to this report offer helpful input into how the introduction of newborn screening for SMA can be implemented across the UK.

Liz Twist MP

Chair of the All-Party Parliamentary Group for Rare, Genetic and Undiagnosed Conditions

Bob Doris MSP

Convener of the Scottish Cross-Party Group on Rare, Genetic and Undiagnosed Conditions

Russell George MS

Chair of the Health and Social Care Committee

Introduction

The UK has established itself as a world leader in science and research, with an increasing number of healthcare and life sciences innovations being developed for the range of conditions affecting the health and prosperity of society, including rare diseases.

With the Government setting out ambitious targets for the UK to be a life sciences superpower and remaining at the forefront of global research and innovation, it is vital that action is taken to ensure that innovative treatment options are reaching those that need them, at the right time. This is even more important for those affected by rare conditions, where early diagnosis and intervention offer significant opportunities to transform outcomes.

Spinal Muscular Atrophy (SMA) is one such condition - a rare, neuromuscular genetic disease that has a devastating impact on those affected and their families.[1] Without treatment, it is the leading genetic cause of infant death in the UK, affecting approximately 70 babies every year, with most of them not surviving past their second birthday without requiring permanent ventilation.[2,3,4]

Early diagnosis of SMA and accompanying treatment provides the strongest opportunity to prevent disease progression and give children with the disease the best chance of a healthy life.[5] Newborn screening is the fastest route to early and pre-symptomatic diagnosis, however, it is not currently included within the Newborn Screening Programme. In contrast, SMA is routinely screened for within more than half of European Union member states, and nearly two-thirds of Europe geographically, including within Russia, Turkey and Ukraine.[6]

The UK National Screening Committee – which is responsible for advising Ministers across the four UK nations on all aspects of screening and programme implementation – has committed to reviewing the case of newborn SMA in light of recent developments in the availability of new treatment options.

This report has been developed in recognition of this review, drawing on input from a range of experts from across the SMA and rare disease community as part of the *Every Moment Matters* campaign, which aims to highlight the urgent need to introduce newborn screening for SMA and the need to utilise the wealth of real-world and international evidence that supports its implementation. This includes shining a spotlight on the real-life impact of SMA on newborns and their families, and the opportunity to transform the lives and outcomes through the introduction of newborn screening.

Urgent action is needed to ensure that newborn screening for SMA is implemented across all four nations in the UK.

> “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**

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We are grateful to Giles Lomax (Chief Executive, SMA UK), Portia Thorman (Advocacy Lead, SMA UK), Professor Felicity Boardman (Deputy Head of the Unit of Social Science and Systems in Health, University of Warwick) and Dr Corinna Clark (Assistant Professor, University of Warwick) for contributing their time and expertise to the development of this report.

We also extend particular thanks to the several families that have shared their experiences of living with the impacts of SMA – these contributions have been invaluable to the development of the report and we are incredibly grateful to those who have shared their stories.

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Chapter 1

What is spinal muscular atrophy?

Spinal Muscular Atrophy (SMA) is a rare, genetic neurodegenerative condition characterised by the progressive loss of motor neurones in the spinal cord.[7,8] It is the most common genetic cause of death in infants.[3] For those with later-onset types of SMA, most live into adolescence or adulthood.[8,9]

There are five types of SMA, which vary in terms of severity and age of onset. Sub-types that appear prenatally or during early infancy are the most severe, resulting in the most rapid loss of motor function and shortest life expectancy.

- **SMA Type 0:** The most severe and rarest form of SMA and onsets in pre-natal infants. Clinical presentations of SMA0 include severe joint deformity, congenital heart defects and as a result, those affected rarely live beyond the first three months of birth.[10]
- **SMA Type 1:** The most common sub-type of SMA, accounting for approximately 60% of all affected infants who are born with SMA.[8] Babies with SMA1 often develop symptoms within the first 6 months, by which time, up to 95% of motor neurones will have been lost.[11] Motor neurons cannot be regenerated, and as a result, will lead to irreversible disability, including respiratory weakness, inability to control limb function and inability to sit independently. As a result, 90% of those with SMA1 do not reach their second birthday or will need permanent ventilation.[4]
- **SMA Type 2:** Accounts for around 20% of all SMA cases and is characterised by infants experiencing delayed symptom onset. For SMA2 the first signs of the condition are visible between 6 and 18 months.[12] Over 90% of infants affected by SMA[2] can be expected to live into their twenties and over half into their forties, however may never be able to stand or move around independently.[9,13]
- **SMA Type 3 and 4:** Characterised by delayed symptom onset, with those affected generally presenting with clinical symptoms of SMA between the ages of 3 and 30.[14] Those with SMA3 and SMA4 can typically sit and move around independently, but can suffer from muscular weakness, respiratory issues and may require regular medical assistance and care, with life expectancy being comparable to those with Type 2 SMA.[9,13,15]

SMA and the SMN Gene

In most cases, a child can only be born with SMA if both parents are carriers of the pathogenic (or 'faulty') SMN gene variant. Parents often don't have SMA themselves, but around 1 in every 40 to 60 people carry the main faulty gene that causes SMA. As such, if both parents carry the SMN gene, there is a 25% chance their child will have SMA, a 50% chance their child will carry the faulty gene but not have SMA themselves, and a 25% chance their child will not carry the faulty gene and will not have SMA.[16]

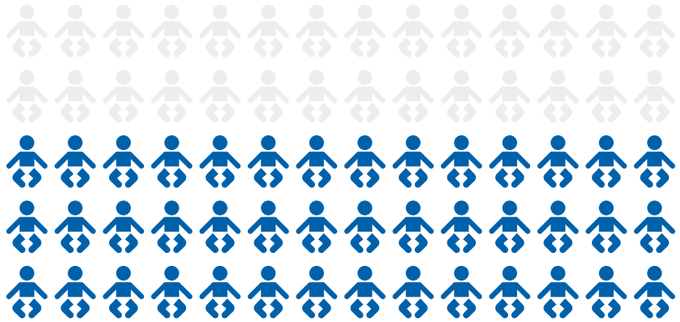
How many people are affected by SMA?

It is estimated that between

1 in 40 and **1 in 60**

are carriers for the defective SMA gene, meaning the number of SMA cases is estimated to be around

1 in 10,000[16,17]



70 babies

are estimated to have been born with SMA in the UK in 2021 and of these

42 would have been expected to be born with SMA1, **the most severe form.**[2]

It is believed that there could be up to

1,340 people

across England, Scotland, Wales and Northern Ireland living with SMA – with some estimates as high as

2,500

- but as there is this no central patient registry, the exact number of those affected is not known.[2,8]

Ezra's Story

Ezra, born in November 2016 to parents Portia and Dan - who had no history of genetic conditions in either family - is one of the first generation of SMA Type 1 children to make it beyond the earliest years of infancy.

Just one month after his birth, Ezra caught a cold. After a week spent in the local hospital, Ezra was admitted to Paediatric Intensive Care (PICU) in London in respiratory distress and required a ventilator to keep him alive.

After his first Christmas was spent in PICU, Ezra was successfully taken off life support, although a PICU nurse reported back to the consultants that she was concerned about his very low muscle tone; he didn't seem to be moving very much at all. This was not investigated any further.

In the new year he was well enough to breathe by himself and was discharged from hospital. At home, Portia and Dan noticed a severe deterioration in Ezra's physical condition, especially his motor skills and his swallowing. He couldn't move his legs or bear any weight; he couldn't hold the weight of his own head or lift his dummy to his mouth; and he started choking on his milk. Portia knew something was not right, but three separate health visitors put his condition down to his recent illness; he even passed his very delayed eight-week check with the GP because he was so bright and alert cognitively.

Finally, in March 2017, when Ezra was 4 months old, the family received the devastating diagnosis of SMA Type 1, a very severe, fast progressing form of the disease. They were told he would probably die before he was one year old.

Thankfully, Ezra could receive treatment through what is known as an Expanded Access Programme on compassionate grounds. In May 2017, he received his first dose. By this point he could only move his forearms. Portia had to give up her teaching career to care for him.

Over the next 3 years Ezra had ten long admissions to PICU where he was intubated and ventilated. Now, aged 6, he has NHS funded 12-hour nursing care overnight and 8 hours of 1:1 nursing at his mainstream school. He drives a powered wheelchair and needs support of a specialist spinal brace to sit up. He is reliant on a biPAP ventilator when he sleeps. He cannot eat or drink orally. He has respiratory physiotherapy sessions everyday using a cough assist machine and suctioning machine to keep his airways and his lungs clear of secretions.

If there had been newborn screening for SMA when Ezra was born, his life and the life of his family would look very different now.

Ollie's Story

Ollie was born in November 2020, at the start of the second UK-wide COVID lockdown. He is the first-born child to Ben and Amy, who had a low-risk pregnancy and straightforward labour. Yet, Ollie was born with SMA Type 1 and the symptoms were there from day one, but they weren't diagnosed at the time.

Early on, Amy had a video-call with a midwife (home visits were restricted due to COVID) where she raised concerns over the way Ollie was breathing from his tummy – intermittently struggling with some breaths – but was told this was normal. The new parents thought nothing more of it.

At five days old the heel prick test was done, and Ollie was weighed; he'd dropped in weight by 11% since birth and so he was kept under midwife review for another six weeks, but no further concerns were raised. Ollie found feeding difficult; looking back, it's clear he was struggling to swallow.

At the six-week GP review, a Moro Reflex test was repeated several times as Ollie wasn't reacting – his arms and legs remained limp. Yet, this was brushed off with "some children don't react, and he appears happy and alert".

By January it was getting really difficult to feed Ollie. After several more appointments, a specialist lactation consultant referred Ollie to a paediatrician with a suspected tongue-tie. It was only at the preoperative assessment that Ollie's low muscle tone and abnormal breathing were highlighted. The paediatrician said he would organise a separate consultation for genetic tests, with a routine follow-up appointment in three months' time. Ben and Amy found the lack of urgency shocking -if they had waited that long, Ollie likely wouldn't have survived.

Ben and Amy became really concerned in late January during a Zoom call with NCT friends. The other babies were moving so much more compared to Ollie and they decided to take him to A&E. There, the registrar shared concerns and admitted Ollie for further tests. This was the first time that any medic had taken Ollie's symptoms seriously – at 11-weeks old he had been consistently assessed by health professionals.

In mid-February, Ben and Amy received the heart-breaking diagnosis that Ollie has SMA Type 1. At three-months' old, Ollie began treatment.

Despite this, in his first two years, Ollie had four PICU admissions on invasive ventilation to get him through colds and viruses. He now requires significant medical support: he sleeps on BiPAP ventilation, is fed through a percutaneous endoscopic gastrostomy (PEG) tube, requires extensive physiotherapy, and twice-daily chest clearance. He also requires night nurses to care for him whilst his parents sleep, and 1:1 nursing at preschool too.

Newborn screening would have made a huge difference to greatly improve his life quality.

Chapter 2:

The impact of SMA

Health and care impacts

For newborns diagnosed with SMA Type 1 (the most common form of the condition), many experience significant and irreversible disability as a result of delayed diagnoses. In the absence of newborn screening, most babies with SMA Type 1 are diagnosed - around the age of 6.3 months, by which time 95% of motor neurons will have been lost.[11] The motor neurones most affected by this condition are those that allow walking, crawling, arm movement, head and neck movement, swallowing and breathing.[8] This means that many babies will miss normal milestones in their first months of life.

Even when treatment is initiated once symptoms have appeared, most of these infants will never walk independently and many will need mechanical ventilation, nutritional support and 24/7 care. This clearly has an enormous impact on children and families directly affected, while also generating significant, and potentially avoidable costs for the NHS and society. Conversely, when treatment is given before a baby shows any symptoms – something made possible through newborn screening – those children could experience a life without severe and progressive weakness.[18]

As a result of these delays, around 90% of babies with SMA Type 1 do not reach their second birthday without permanent ventilation, while most of those diagnosed with SMA Type 2 will never walk independently.[4,13]

In addition to the immediate physical impacts of SMA, the condition also generates considerable, multi-faceted emotional and economic burdens.

Surveys conducted by SMA UK have sought to understand how SMA impacts those affected and their caregivers' lives in more detail. Responses to two surveys – which SMA UK estimate represents between 10 and 15% of the total UK SMA population – highlight the significant impact of the condition on mobility – particularly on ability to sit without support, roll independently and stand/walk unaided.[19] Those with SMA similarly have high levels of need for specialist equipment and other more specialist health interventions – many of which are not funded by the NHS – as well as regular contact with healthcare professionals (HCPs). Almost one third of people with SMA see more than 10 HCPs per year.[19]

Caregivers also spend a significant number of hours per week caring for someone with SMA. This varies based on the type of SMA and level of disability experienced, but for those unable to sit without support (a clinical indicator of SMA Type 1), carers report spending an average of 117 hours per week providing care. On average, almost three unpaid caregivers provide support to someone with SMA Type 1 and Type 2 – and can include parents, partners, siblings, grandparents, relatives, friends and neighbours.[19]

Studies have also demonstrated that those caring for someone with SMA Type 1 are three times more likely to spend an excess of 10 hours per day on care, compared to those caring for someone with SMA Type 3, which presents at a later stage and is less severe clinical presentation.[20]

For many carers the most significant impact is on their jobs and careers. For those caring for a child with SMA Type 1, 74% report that one or more caregivers had given up work completely, while others have refused promotions or changed career goals as a result of their carer responsibilities. These impacts are also significant but lower for those sharing for someone with SMA Type 2 or Type 3.[19]

[i] n = 125 for survey A and n = 188 for survey B

Financial Impacts

In light of the impacts outlined above, SMA also places a substantial financial burden on society – both in terms of direct costs to the healthcare system, as well as indirect costs as a result of lost productivity.

Estimates of the impact of SMA on the NHS vary by type and study. SMA UK has estimated for instance that on average, a person with SMA generates costs to the NHS of £49,723 each year due to the associated interventions, treatments and formal care needs – however this does not take into account the additional costs associated with hospital admissions and high-dependency units.[19]

In a study of 86 children with SMA – 26.7% having SMA Type 1 and 73.3% having SMA Type 2 and 3 - the annual average cost reaches €54,295 in the UK. The direct non-healthcare costs ranged between 79–86% of the total cost and the informal care costs were the main component of these costs.[21]

Further studies have indicated that children with SMA Type 1 receiving treatment spent a median of 118 days in hospital in the 24 months following their diagnosis.[22] According to the study, children with SMA Type 1 occupied 17% of the total High Dependency Unit (HDU) capacity and 4.2% of the Paediatric Intensive Care Unit (PICU) capacity in the region. This incurred costs of £1,626 per day for an HDU bed and £1,785 to £3,784 for a PICU bed. In total, children with SMA Type 1 spent a median (range) of 20% of their life in hospital, with an estimated cost of this care being £2.2 million.[22]

Other studies have estimated the total costs associated with each annual cohort of newborns with SMA based on the absence of newborn screening in the UK, which came to £222,259,604 (compared to £160,086,073 had newborn screening been available). This therefore represents a potential incremental cost savings of £62,191,531 for the NHS, for each annual cohort of newborns with SMA.[23]

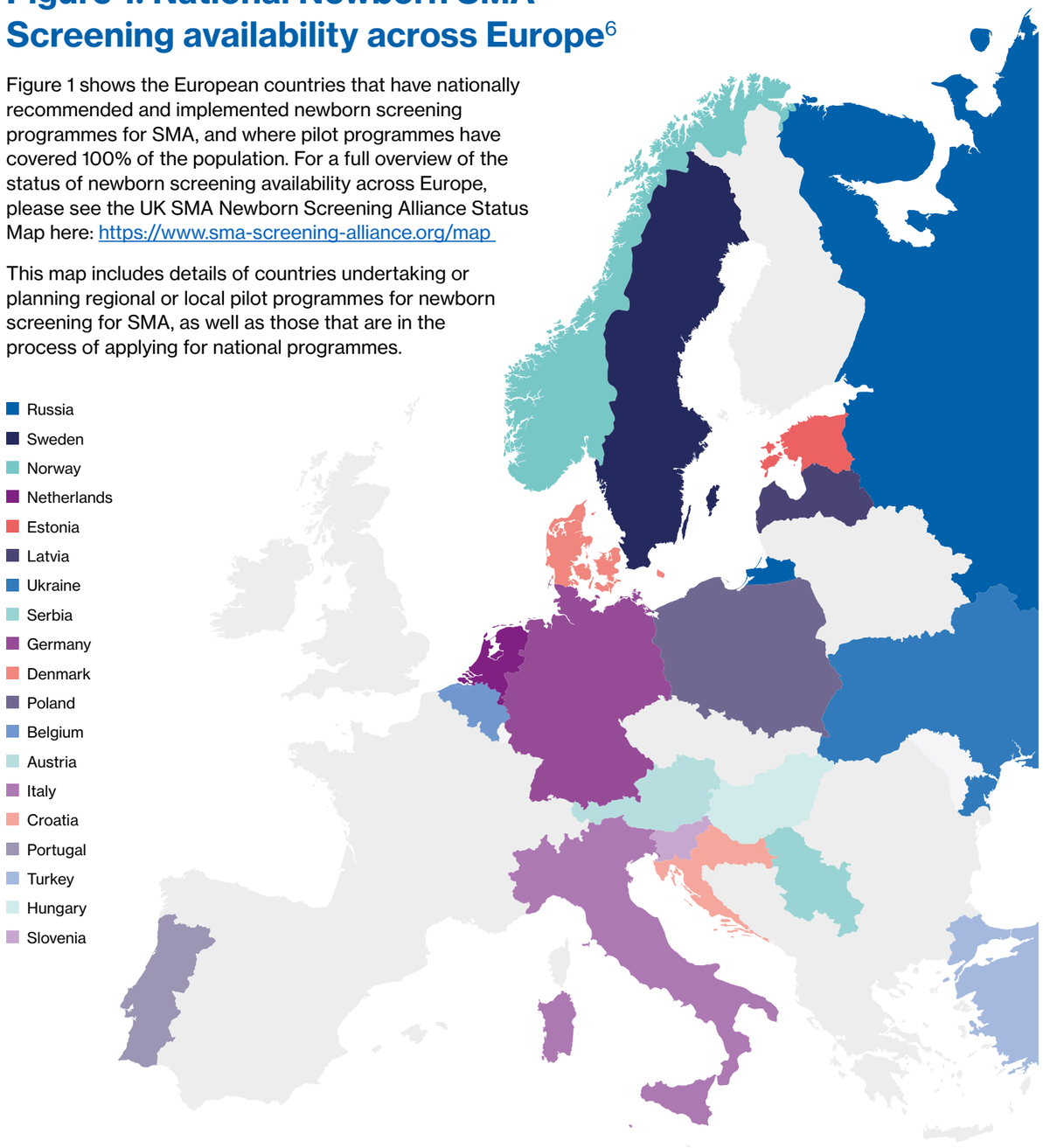
Financial impacts are similarly seen as a result of productivity losses among parents, families and friends. For example, it has been estimated that the average annual cost of lost productivity per unpaid caregivers amounts to £14,350 based on reducing their working hours by 25 hours per week.[19] Carers also incur significant out of pocket (OOP) costs for health materials, travel, and accommodation – on average £8,025 per person per year – however this does not include any costs of care that may be paid for by caregivers.[19]

SMA clearly generates significant burdens on the healthcare system and on the lives of those affected, however opportunities exist to facilitate earlier intervention that can substantially reduce these impacts.

Figure 1: National Newborn SMA Screening availability across Europe[6]

Figure 1 shows the European countries that have nationally recommended and implemented newborn screening programmes for SMA, and where pilot programmes have covered 100% of the population. For a full overview of the status of newborn screening availability across Europe, please see the UK SMA Newborn Screening Alliance Status Map here: https://www.sma-screening-alliance.org/map

This map includes details of countries undertaking or planning regional or local pilot programmes for newborn screening for SMA, as well as those that are in the process of applying for national programmes.



1. Russia (All newborns screened since January 2023) 2. Sweden (All newborns screened since August 2023) 3. Norway (All newborns screened since 2022) 4. Netherlands (All newborns screened since 2022) 5. Estonia (All newborns screened since July 2022 via pilot programme) 6. Latvia (All newborns screened since April 2023, and since February 2021 via a pilot programme) 7. Ukraine (Screening ongoing in areas under effective Ukrainian control; start dates vary by region) 8. Serbia (All newborns screened since September 2023) 9. Germany (All newborns screened since July 2021) 10. Denmark (All newborns screened since January 2023) 11. Poland (All newborns screened since March 2022) 12. Belgium (All newborns screened since March 2022) 13. Austria (All newborns screened since June 2021) 14. Italy (Screening for SMA in most of the 20 regions) 15. Croatia (All newborns screened since March 2023) 16. Portugal (All newborns screened since October 2022) 17. Turkey (All newborns screened since March 2022) 18. Hungary (Majority newborns screened since November 2022) 19. Slovenia (Recommended in November 2022)

Chapter 3:
Current pathways and management

Diagnosis

For parents with a known family history of SMA, genetic blood testing is used to determine whether either is a carrier of the faulty gene that can cause SMA. Testing for the disease can also be conducted prenatally and is carried out if both parents are identified as carriers following genetic testing or if developmental abnormalities are identified during pregnancy.

For parents that do not have a known family history of SMA – and therefore are not known to be carriers of the faulty gene – diagnoses are often made after children start to develop symptoms, which is generally within the first six months of life. On average this means that most babies with SMA Type 1 are diagnosed around the age of 6.3 months, by which time 95% of motor neurons will have been lost.[11]

As a result of these differences, significant inequalities exist between families with a known family history versus those without, as the latter are reliant on symptom recognition among healthcare professionals in order to receive a diagnosis. As treatment for SMA cannot reverse disability but can slow progression, diagnosis delays contribute to inequalities in outcomes experienced amongst those who are not ultimately able to receive pre-symptomatic treatment.

> “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[11]

Management and treatment

Management and treatment of SMA can depend on the disease type and symptoms, as well as response to therapies administered, which can vary from child to child. As a result of motor neuron loss, children will likely be severely impacted by varying degrees of muscle weakness, affecting their potential to meet key early-life development milestones, such as the ability to crawl and walk; their arm, hand, head and neck movement; and the ability to breathe and swallow.[1] Infants who survive will typically require significant care and support as a result.

For babies and children with diagnosed SMA, care should be delivered by a multi-disciplinary team (MDT) that may include a combination of specialists in paediatric neuromuscular conditions, respiratory care, physiotherapy, occupational therapy, dietetics and nutritional care, speech and language therapy, palliative care and community care.[24] While SMA cannot be cured, the aim of the MDT is to manage symptoms, reduce complications associated with muscle weakness and maintain quality of life.

Newborns and children with SMA Type 1 respiratory problems are common – particularly for those unable to sit - and mechanical breathing support (respiratory life support) may therefore be required, alongside regulatory respiratory check-ups and overnight sleep studies to assess overnight breathing.[24]

Support for feeding and swallowing may also be needed, with the 2017 Standards of Care (SoC) recommending all children who are unable to sit should have a swallow study shortly after diagnosis and again if they show other signs of difficulties. Where children have difficulties with feeding, tube feeding may be required – with some children needed gastrostomy (PEG) tubes as a long-term solution.[24]

Without 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.[4,13]

Evelyn's Story

Evelyn was born in November 2020, without complication, and appeared a healthy, happy and beautiful baby. She passed her 8-week GP check with flying colours and was seen by several health visitors in her early months, who felt she was developing well.

At 4 months old, it was noticed by her mother when holding another child for the first time since the pandemic lockdown, that by comparison Evelyn lacked muscle tone and had limited movement. She also had not attempted to start rolling over and was struggling to support her own head. Alarm bells started to ring for her mother after she had researched information online and realised certain milestones had also been missed. Her GP was called, but felt there was nothing to worry about and no urgency for her to be examined.

Evelyn was diagnosed with SMA type 1, aged 5 months old, after finally being seen in A&E and referred for genetic blood tests. The family were referred to a paediatric neurologist, and treatment options were subsequently discussed.

Evelyn's disability continued to progress post-infusion, and by the age of 10 months, she required non-invasive ventilation for up to 16 hours a day. She was unable to sit unaided, had very limited movement, required 24-hour monitoring, regular suctioning, and was tube fed with a pump. Her mother was unable to return to work and became a full-time carer. Evelyn's father was signed off at work through stress, and her older sister began to struggle with anxiety. Small signs of improvement did appear by 12-13 months of age, but this was limited to the peripheral areas of her body only.

Aged 16 months old, Evelyn was hospitalised again for a medical concern unrelated to her SMA. In a heartbreaking turn of events, she was found to be suffering with hydrocephalus, caused by an unidentified mass in her lower spine. Taking into account her significant respiratory distress and deteriorating SMA-related symptoms, it was decided by Evelyn's multi-disciplinary team, that she was not healthy enough to withstand the surgery required. She was subsequently moved into palliative care at a local children's hospice to make her as comfortable as possible. Evelyn bravely battled for 6 weeks, before passing away on 5th April 2022 with her parents by her side.

Evelyn's postmortem results showed that she had retained minimal motor neurones, and her treatment had been given too late to have a marked effect.

Chapter 4:

Opportunities to transform the lives of children and their families

Despite the availability of effective treatment options that can transform the impact of SMA when delivered pre-symptomatically, around 70 babies are still born in the UK each year which face suffering irreversible damage due to the absence of newborn screening for the condition, and subsequent associated delays in accessing treatment.[2]

Early diagnosis of SMA through newborn screening, and accompanying treatment, therefore provides the strongest opportunity to prevent disease progression and give children with SMA the best chance of a healthy life.[5]

In the UK, the Newborn Screening Programme (NBS) is a national programme which systemically screens newborn babies for rare diseases in England through the heel-prick blood spot test, allowing for early disease identification and timely intervention. The conditions included within the NBS are informed by recommendations made by the UK-wide National Screening Committee (UK NSC), which currently recommends newborn screening for nine rare but serious conditions. This includes: sickle cell disease, cystic fibrosis, congenital hypothyroidism, phenylketonuria, medium-chain acyl-CoA dehydrogenase deficiency, maple syrup urine disease, isovaleric acidaemia, glutaric aciduria type 1 and homocystinuria.[25]

SMA is not currently included as one of the conditions in the Newborn Screening Programme. It is, however, screened for within more than half of European Union member states, and nearly two-thirds of Europe geographically, including countries such as Russia, Turkey and Ukraine.[6] As a result, the SMA Newborn Screening Alliance in Europe has estimated that by the end of 2022, 40% of newborns in continental Europe were screened for SMA.[26]

Economic modelling estimates that the introduction of newborn screening for SMA within the UK followed by treatment could lead to associated lifetime savings of over £62,191,531 for each annual cohort of newborns (approximately 585,000) identified with the disease, as well as 529 quality adjusted life years (QALYs), compared to a pathway without newborn screening. Evidence suggests that an SMA treatment pathway that starts with screening is also less costly to the NHS.[23]

In countries that already screen newborns for severe combined immunodeficiency (SCID) – a rare group of inherited genetic disorders – there are further cost saving opportunities as the same testing equipment can be used for newborn screening of SMA. Research has shown that the combined introduction of newborn screening for SMA and SCID is cost-effective in the longer term, saving lives, improving outcomes and reducing healthcare utilisation. In the US for instance, the combined introduction of SCID and SMA into the newborn screening panel, alongside early treatment of screen-detected SMA by gene therapy and SCID by early HSCT, is projected to result in $8.6 million in cost savings and 95 QALYs / 137 Life-Years (LYs) gained, per 100,000 newborns, over a 60-year time horizon.[27]

Newborn screening for SCID is currently under review by the UK NSC, with an in-service evaluation currently underway in England and covering two thirds of the population, expected to finish in March 2024.[28]

Giles Lomax, Chief Executive, SMA UK

"In 2018, the UK Newborn Screening Committee (UK NSC) unanimously rejected the opportunity for SMA to be included on the existing blood spot test. We know that since that rejection, over 300 babies have been born with SMA, that's one child every five days, that will likely not be diagnosed until irreversible muscle atrophy has happened.

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.

We understand the importance of considering as much robust evidence as possible to inform the NSC's eventual decision on whether to recommend routine newborn screening for SMA across the UK including 'to do more good than harm at reasonable cost'. However, we believe that the balance is very much in favour of SMA being included on the newborn screening programme and the need for speed is critical if we are to prevent unnecessary devastation for families watching their child lose the ability to move, breathe and eat independently.

From countless testimonies and examples, we know that early intervention is key through the disease modifying pioneering treatments available. We are seeing children who are treated early enough, going on to live very normal lives with minimal medical interventions related to SMA, these are incredible outcomes when compared to the natural history of SMA.

Along with our partners, we will work tirelessly to ensure that SMA is added onto the UK NBS programme."

Chapter 5:

Newborn SMA Screening in the UK

The UK National Screening Committee's review of SMA

In 2018, the UK NSC issued a negative recommendation on widespread screening of SMA, citing several contributory factors at the time, including a lack of information on the number of people affected; limited evidence about the acceptability and reliability of screening; and a lack of effective treatments for the condition.[29] In the following years however, new treatment approaches have since become available and many of these perceived barriers to screening have since been addressed.

In November 2022, the UK NSC confirmed it would review the case for newborn SMA screening based on significant developments, such as the availability of new treatment options.[30] In June 2023, the UK NSC announced following its June 2023 meeting that it will be commissioning a new 'comprehensive and flexible' cost effectiveness modelling study, and that it will simultaneously start scoping an in-service evaluation (ISE) for SMA newborn screening.[31] This decision to work on these elements of the review in tandem - rather than sequentially – has been broadly welcomed by the stakeholder community and demonstrates the opportunities that exist to expedite UK NSC processes where there is significant and urgent need.

Despite this, there are concerns around the size, scale and scope of the proposed ISE, as well as the timelines associated with its implementation and final read-out. The delivery of a regional pilot for example risks creating inequalities between newborns inside and outside of the scope of the programme, including within England and between devolved nations.

Some recent ISE programmes lead by the UK NSC – for example for severe combined immunodeficiency (SCID) - are anticipated to take over seven years to deliver from the point at which they were first announced, which if replicated, would see newborn screening for SMA not made routinely available until 2030, on the basis of the SMA ISE commencing in 2024.[28] Not only must these lengthy timelines not be repeated for SMA, but it is also imperative that any other UK NSC ISEs workstreams do not impact the timely implementation and delivery of the ISE for SMA.

Similarly, ISEs previously run by the UK NSC have historically been delivered geographically within just England, as funding is derived from the England-based National Institute for Health and Care Research (NIHR). If repeated for the SMA ISE, this could lead to a further widening of inequalities between those affected in England, Scotland, Wales and Northern Ireland.

Ultimately, the longer the review timelines are, the more the UK will continue to lag behind other comparable health systems, and the greater the number of newborns and families who will ultimately experience barriers to timely diagnosis and access to pre-symptomatic treatment that has the ability to transform the life prospects of babies born with this condition.

Extended timelines could similarly result in misalignment across different parts of the system, creating an environment where patients are unable to access treatment options at the point when they are most effective, due to avoidable delays in diagnosis, leading to worse outcomes and additional healthcare utilisation than if screening were available, as well as also running counter to national policy ambitions of addressing the diagnostic odyssey experienced by people with rare diseases.[23,32]

The Human Cost of Delaying Newborn SMA Screening

Since the first treatments were made routinely available to children with SMA on the NHS on 1 June 2019, the UK SMA Newborn Screening Alliance's Monthly Barometer – which has been developed to demonstrate the human cost of delaying newborn SMA screening in the UK – estimates that as of July 2023, 315 babies have been born with SMA since treatment has been made available. This means that 315 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.[33]

Considerations for the introduction of newborn screening for rare diseases

Stakeholders from across the rare disease community have highlighted the opportunity to expand the UK's newborn screening programme to improve early diagnosis of a broader range of rare and genetic conditions, and bring the UK in line with comparator countries across the globe that already screen for conditions such as SMA. However, there are specific challenges relating to the assessment of rare conditions for newborn screening owing to small patient numbers and inherent uncertainties in evidence.

The All-Party Parliamentary Groups for Muscular Dystrophy and Rare, Genetic and Undiagnosed Conditions coordinated an inquiry into newborn screening for rare conditions and published a report in May 2023. A key element of this inquiry sought to understand the types of evidence that should be considered by the UK NSC to allow robust but timely decision-making, and how uncertainty arising from evidence relating to rare conditions should be handled.[34]

Based on 46 evidence sessions received from stakeholders in the rare disease community, the APPG concluded that a review of UK NSC criteria is needed to ensure evidence requirements are appropriate for rare conditions, recommending that approaches to evidence requirements should align more closely to those used by regulators and health technology assessment bodies. This is intended to not only avoid duplication in evidence assessments, but also reduce the burden placed on rare condition stakeholders to produce evidence for multiple different assessments of the same condition. The APPG similarly called for greater consideration of real-world evidence, such as data about treatments collected via Managed Access Programmes which demonstrate the clinical effectiveness and impact of intervention – to support the timely review of rare conditions being considered for newborn screening.[34]

The Genetic Alliance's Patient Charter on Newborn Screening also made the following recommendations on adapting decision making to fit rare diseases:

1. The methodology for decision-making on newborn screening should be adapted in recognition that the conditions being screening for are rare and thus present significant challenges;
2. Decisions on newborn screening should be made by a body with specific and relevant expertise;
3. Benefits to the patient, family and broader society other than preventative interventions should also be considered.[35]

> "The UK has not followed the majority of high-income countries in embracing the broad value of newborn screening. Very few high-income countries screen for as few conditions as the UK."
>
> **Genetic Alliance: Fixing the present, Building for the future – Newborn screening for rare conditions report (2020)[35]**

EURORDIS Key Principles for Newborn Screening

EURORDIS-Rare Disease Europe is a non-profit alliance of over 1,000 rare disease patient organisations from 74 countries that work together to improve the lives of over 300 million people living with a rare disease globally.[36]

In recognition of the *"marked variation in how NBS is conducted within and between countries and in the coverage of NBS programmes"* EURORDIS set up the Newborn Screening Working Group to define 11 key principles that can be adopted at a national level in order to frame policy discussions and developments of NBS Screening Programmes.[37] These principles are:

1. Screening should identify opportunities to help the newborn and the family as broadly as possible. That is, screening should identify actionable diseases including treatable diseases.
2. NBS should be organised as a system with clearly defined roles, responsibilities, accountability and communication pathways that are embedded into the national health care system and recognised as a mechanism for earlier diagnosis of actionable conditions as part of the broader care pathway.
3. The family of the newborn who has been diagnosed through NBS should be provided with psychological, social and economic support by the competent national healthcare authorities.
4. All stakeholders should be included in the different stages of the NBS process.
5. Transparent and robust governance for expanding NBS programmes is needed. Every country/region should have a clearly defined transparent, independent, impartial and evidence-based process for deciding which conditions are covered by the NBS programme that includes all stakeholders.
6. Governance of NBS programmes should be explicit, comprehensive, transparent and accountable to national authorities.
7. The evaluation process on the inclusion/exclusion of diseases in NBS programmes needs to be based on the best available evidence, reflecting health economic evidence but not determined only by health economics.
8. Information and education of all stakeholders on rare diseases and the whole NBS process is essential for a broad and fair implementation of NBS programmes.
9. European-wide standards addressing the timing, sample collection methods, follow-up, and information shared with parents are needed to guarantee quality throughout the process.
10. Blood spot samples should be stored in national biobanks for quality control and research purposes while ensuring appropriate measures for data access as well as robust safeguards for data protection and privacy are in place.
11. ERN affiliated centres should be integrated in the care pathways of the different Healthcare systems and should be considered as preferential partners in providing recommendations on NBS policies.

SMA demonstrates significant opportunity for the UK to align with these internationally developed principles as an 'actionable disease' – whereby early invention leads to health gains for the newborn, early diagnosis avoids the lengthy 'diagnostic odyssey, and parents will have reproductive options during subsequent pregnancies.

The impact of the UK NSC on newborns in Scotland, Wales and Northern Ireland

While delivery of health and care services are the responsibility of devolved administrations, the National Screening Committee has a UK-wide remit and is the sole body responsible for making screening recommendations to Ministers in Scotland, Wales and Northern Ireland. Once the UK NSC makes a recommendation it is up to individual healthcare systems in the devolved nations to pull together an implementation plan, and experience has shown that the implementation of UKNSC advice can vary depending on the level of system readiness. For example, Northern Ireland was, until recently, an outlier in that it only screened for five conditions before reaching parity with the rest of the UK.

At present, regional pilot programmes are proposed and underway to assess the effectiveness of newborn screening for SMA across the UK. In England, the University of Oxford announced it would be coordinating the first UK pilot study of newborn screening for SMA in the Thames Valley – lead by Professor Laurent Servais.[11] In Scotland, a pilot programme is under consideration while the UK NSC undertakes its review – lead by Professor Zosia Miedzybrodzka, Clinical Lead for the Scottish Genomics Network.[31] However, both Wales and Northern Ireland are absent from either the established pilot in the Thames Valley or the proposed pilot in Scotland.

The recently announced ISE for SMA therefore presents an important opportunity to ensure that inequalities in access to newborn screening across the UK are not deepened by partial or regional coverage. This is particularly important for newborns in Wales and Northern Ireland, where there are currently no regional-level pilot programmes.

One key criteria for the introduction of a new condition to the screening list is the routine availability of an effective treatment. Given that Scotland and England have separate bodies responsible for the introduction of new medicines to the NHS, this can often result in variable patient access. This was recently true for SMA where Scotland had approved therapies for the pre-symptomatic treatment of SMA, whereas England had not. However, despite this pre-symptomatic approval, there was no clear means of identifying and treating these patients without a screening programme in place. This particular inequity was raised by clinicians in the Scottish Medical Journal.

> “NBS does not come without important ethical, societal and financial implications. There are myriad reasons why NBS is not suitable for many genetic conditions at present. However, as professionals involved in SMA research, diagnosis and patient care in Scotland, we believe that all of the necessary data, evidence, and therapeutic approvals are in place to justify the inclusion of SMA on NBS programmes in Scotland.”

A call to introduce newborn screening for SMA in Scotland, published in the Scottish Medical Journal[38]

The role of newborn screening in achieving the UK’s rare disease and life science policy ambitions

In addition to clear health and economic arguments for introducing newborn screening for SMA across the UK, its implementation would also support wider existing policy ambitions. The UK Rare Disease Framework highlights the importance of achieving faster final diagnosis, helping to address the ‘diagnostic odyssey’ faced by many with rare diseases.[32]

All four nations have published subsequent Rare Disease Action Plans which highlight the importance of early diagnosis for patients with rare diseases and the role of newborn screening within this. The Rare Disease Action Plan for England for example includes commitments to encourage greater collaboration between the UK NSC, researchers and stakeholders, and to identify practical approaches to developing evidence that can help the Committee make *“rapid and robust”* decisions about newborn screening for rare diseases.[39] Scotland’s Rare Disease Action Plan similarly contains commitments to engage with any new UK-wide screening research pilots, while the Northern Ireland Rare Disease Action Plan notes plans to review newborn screening, participate in the UK NSC and engage with UK-wide screening initiatives.[40,41] The Rare Disease Plan for Wales similarly emphasises the importance of establishing a public health and screening system in Wales that uses genomics to strengthen the current biochemical screening, diagnostic and care pathways in those at high risk.[42]

The Government’s Life Sciences Vision sets out that the UK aspires to be a world leader for development, testing, access and uptake of new and innovative treatments and technologies.[43] Timely implementation of newborn screening is a critical element of realising this ambition and providing access to potentially life-saving treatment. Screening for SMA similarly provides a tangible example of how barriers to innovation can be addressed, and more broadly how the UK’s life sciences ambitions should be translated into supporting the best possible outcomes for newborns and families across the UK.

The role of the Newborn Genomes Programme in the context of SMA

In December 2022, the UK Government and Genomics England launched the Newborn Genomes Programme – a research study intended to explore the effectiveness of using whole genome sequencing (WGS) to detect rare diseases in newborn babies.[44,45]

While the programme offers the significant potential to detect thousands of genetic conditions at birth, and could have a major impact on the quality of life for children born with the conditions in the future, it is important to recognise that this study is in the early stages of research and the benefits of WGS are not due to be realised for several years to come. As such, stakeholders have emphasised that this programme should not overshadow nor undermine the potential of expanding the newborn screening programme in the interim.

It is therefore vital that the UK capitalise on the tools it already has at its disposal to bridge the gap between future innovations such as WGS and the immediate needs of babies born with rare genetic conditions such as SMA today. Newborn screening through the existing bloodspot programme is therefore the best and most tangible solution to delayed diagnosis and avoidable disability and mortality for babies born with SMA.

As the Genetic Alliance has noted, *"This technology may not be as innovative or fashionable as whole genome sequencing, but it has just as great an ability to improve the lives of babies and their families where tests are available for a particular rare condition.*

We don't screen newborns for SMA in the UK, whereas other countries do, and this urgently needs to change." [46]

The acceptability of newborn SMA screening among parents and families

Prof Felicity Boardman and Dr Corinna Clark (Warwick University) are currently collecting data to address the gap in evidence highlighted in the 2018 UK NSC SMA screening review relating to the acceptability of SMA newborn screening. They are conducting a large-scale acceptability study with members of the public, people and families affected by SMA, and health care professionals.

Using surveys with over 10,000 people, and over 30 interviews with representatives from each stakeholder group, they will explore the breadth and depth of views towards screening for SMA. They are looking at a range of ethical considerations, including the impact of an SMA diagnosis on families where their child would not currently be eligible for presymptomatic treatment (which would be the case for babies expected to develop later onset types of SMA).

Data collection and analysis for this study is currently ongoing. Preliminary findings from initial responses to the public survey suggest levels of support for implementing SMA newborn screening at over 80%, with the majority of the remainder being undecided and less than 5% not in support. It is expected that the full results from the surveys and interviews will be available in March 2024.

Linking up with the Thames Valley Screening Pilot, Boardman and Clark are also contacting parents approached to take part in the screening pilot to understand parental decision-making around screening for SMA. This includes the reasons why parents choose to accept, decline, or are undecided about screening for their child. Initial findings from the Thames Valley screening pilot are expected in June 2024.

Recommendations

Contributions to this report and commentary from families and the stakeholder community have made clear that the continued absence of newborn screening for SMA in the UK is unacceptable and is resulting in devastating and avoidable impacts on those affected. The following recommendations are therefore made to support the urgent review and implementation of screening, to ensure that the UK realises the potential to save and improve lives, and reduce the significant healthcare costs associated with current standard of care.

1. The UK National Screening Committee should announce at its earliest convenience 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 as a result of individual and country-specific pilot programmes, and reflecting the UK-wide remit of the UK NSC, 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.

2. The UK Government must ensure sufficient funding is available – and its Barnett formula equivalent - 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.

3. The UK National Screening Committee should set out clear opportunities for collaboration with experts from 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 full system readiness for the adoption of screening and equitable implementation across all four nations.

4. 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.

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