Babies born with Spinal Muscular Atrophy at risk of irreversible, life-changing damage, despite innovative treatments, as the UK lags behind other European countries in newborn screening

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London, UK, 25th October 2023 – Latest research shows the UK lagging behind its European peers in its newborn screening (NBS) programme, down in 23rd place out of over 30 European countries.[1] This is a drop from 18th place in 2022.[2] The UK currently tests for just nine out of a potential 50 severe diseases.[1]

Since March 2021, 13 European countries have added the rare neuromuscular disease Spinal Muscular Atrophy (SMA) to their newborn screening panels, many of them with far smaller GDPs than the UK, including Ukraine, Poland and Croatia.[1,3-5] The latest to start screening for SMA is Sweden, which now routinely tests for 27 diseases.[1] In the UK however, there is a further delay to NBS for SMA as the UK National Screening Committee (UKNSC) have now called for a new pilot study to be commissioned.[6]

Without NBS, a baby born with SMA type 1, unless they have a sibling with the condition, will not be identified until symptoms appear, with the average age of diagnosis ~6 months.[7] By this time, 95% of their neurons are lost, resulting in irreversible damage to the nerves responsible for muscle function, causing difficulties with eating, moving, and breathing.[8,9] These complications could be avoided with early diagnosis and subsequent treatment.[10]

Professor Laurent Servais, Professor of Paediatric Neuromuscular Disease at the University of Oxford, commented; *"We are far behind Ukraine, which introduced newborn screening for SMA in a state of war, and several countries in wider Europe where 65% of newborns are screened for SMA. We need to work hard with the National Screening Committee to help prevent the devastating consequences of SMA by introducing newborn screening in the UK. We are currently missing the chance to provide optimal treatment for one child born with SMA every five days."*

NHS England has applauded how innovative treatments have transformed survival in babies born with SMA, however, unless babies are treated as early as possible, they will miss out on the full benefits of receiving these treatments to improve their quality of life and save NHS funding.[11] NBS would allow prompt diagnosis of SMA soon after birth, meaning appropriate care plans can be put in place before symptoms become visible. Regardless of which treatment is chosen by the treating physician, clinical data have proven that the earlier a patient is diagnosed and treated, the better the chance for successful outcomes.[10]

Giles Lomax, CEO of charity SMA UK commented; *"We see first-hand the difference early diagnosis and treatment makes. Where families have two affected children, one treated after symptoms have progressed and one treated at birth following genetic testing, the differences in the health and mobility of the children are striking. Any delays to initiating a UK-wide pilot, where every newborn in the UK is screened for SMA, has life-changing consequences for babies born with SMA and their families."*

NBS followed by treatment for SMA in England has also been shown to be less costly than a treatment pathway without NBS, in a recent cost-effectiveness analysis, with lifetime savings of over £62 million for each annual cohort of newborns identified and improved health outcomes.[10]

Sophie Abbott, mother of baby Milo who was diagnosed with SMA at five weeks commented; *"As a parent, I know the impact the late diagnosis of SMA has had on our family. It is unethical to further delay adding SMA to the newborn screening panel, as early diagnosis and intervention is vital to give as many children as possible affected by the disease the best chance of a healthy life."*

About Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a rare, genetic neuromuscular disease and a leading genetic cause of infant death.[7,12] Caused by the lack of a functional *SMN1* gene, the most severe forms of SMA results in the rapid and irreversible loss of motor neurons, affecting muscle functions including breathing, swallowing and basic movement.[13] Severity varies across a spectrum of types corresponding to the number of copies of the back-up *SMN2* gene.[14] The majority (>70%) of patients with two copies of *SMN2* develop Type 1, the most common form accounting for 60% of cases.[15,16] Type 1 is severe and, left untreated, leads to death or the need for permanent ventilation by the age of two in more than 90% of cases.[7,12] Most patients (>80%) with three copies of *SMN2* develop Type 2, accounting for 30% of cases.[15] Left untreated, patients with Type 2 are unable to walk and will require a wheelchair, and more than 30% will die by age 25.[17] Loss of motor neurons cannot be reversed, so it is imperative to diagnose SMA and begin treatment, including proactive supportive care, as early as possible to halt irreversible motor neuron loss and disease progression.[18,19]

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