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Newborn Screening for Spinal Muscular Atrophy in the UK

Cost-effectiveness and improving quality of life

Spinal Muscular Atrophy (SMA) is a rare progressive genetic disease characterised by muscle loss. Left untreated, most infants don't reach their **second birthday.**[1,2]

Newborn screening (NBS) can enable early detection and diagnosis of genetic conditions, leading to faster access to lifesaving treatment.[3]

There is no national NBS programme for SMA in the UK, yet a study in England shows NBS followed by treatment for SMA is less costly than a treatment pathway without NBS, with lifetime savings of over **£62 million** for each annual cohort of newborns identified.*[4]

The impact of delay in the clinical diagnosis of SMA

Neurodegeneration in the natural history of untreated SMA Type I

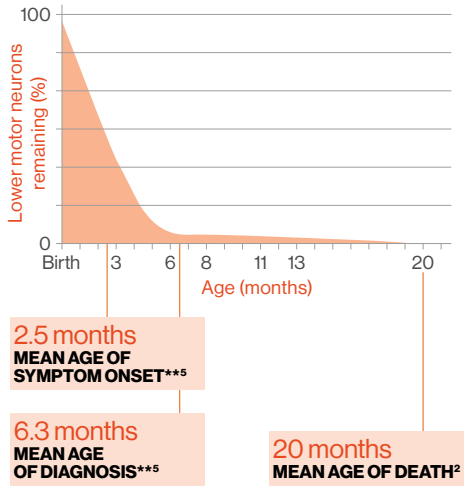

Figure adapted from Mendell JR, *et al.* 2017, and Lin CW, *et al.* 2015.[2,5]

70
NEW CASES A YEAR
In the UK, approximately one child every 5 days is born with SMA, leading to around 70 new cases in the UK each year.[6]

42
CASES OF SMA TYPE 1
Around 42 of these are born with the most severe form of SMA, Type 1.[7]

95%
OF LOWER MOTOR NEURONS LOST BY 6 MONTHS OF AGE
Clinical diagnosis of SMA Type 1 can take over 6 months, during which time 95% of lower motor neurons may be lost.[5,8]

20 months
MEAN AGE OF DEATH
SMA is the leading genetic cause of infant death when left untreated.[1,2]

Where it is available, NBS has improved the possibility of early intervention in SMA. Typically, the time from birth until diagnosis for babies with SMA Type 1 is:

14
DAYS WITH NBS[9]

143
DAYS WITHOUT NBS[10]

Data are from different countries in Europe and so are not directly comparable.

Why does the UK need a national NBS programme?

NBS is the process of systematically testing babies just after birth to identify certain treatable diseases.[11]

Screening tests are quick, simple, and have the potential to detect rare but serious conditions, including progressive genetic diseases.[11]

NBS can enable early diagnosis and timely intervention within the first days or weeks of life before symptoms develop.[3]

Diagnosing and treating babies early is crucial in SMA as neurodegeneration is irreversible.[3]

Pilot programmes in several countries have demonstrated the feasibility of NBS for SMA in Europe.[1,12,13]

- Belgium, Germany, Norway, Poland, and the Netherlands, among others, are actively screening nationwide[12]
- Regional implementation of NBS is ongoing in Spain[12]
- In the UK, regional pilots are still ongoing[12]

65% of children born in Wider Europe (including Russia, Turkey, and South Caucasus) are being routinely screened for SMA.[12]

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A cost-utility analysis found that NBS for SMA in England was less costly and more effective than without NBS*[4]

Lifetime savings of **£62,191,531**

Estimated gain of **529 QALYs**
(**QALY** - Quality-adjusted life year)

NBS followed by presymptomatic treatment results in improved health outcomes for patients with SMA, **and is a cost-effective use of NHS resources.**

From the perspective of NHS England, the analysis compared the costs of:

NBS for SMA in patients either symptomatic or presymptomatic at diagnosis

vs.

Non-NBS patients with SMA who were symptomatic at diagnosis

Costs considered over an SMA patient's lifetime[14-16]

Direct costs: Inpatient, outpatient, and emergency care; medications; medical devices and mobility aids; respiratory and ventilatory assistance; and transportation.

Indirect costs: Loss of productivity, time spent caregiving, anxiety and stress related to caregiving, and changes in employment status for the caregiver.

NBS, newborn screening; NHS, National Health Service; QALY, quality-adjusted life year; SMA, spinal muscular atrophy.

*The cost-utility analysis tested 585,195 newborns and identified approximately 56 with SMA (96% of all SMA patients in England). Cost of each heel-prick screening test assumed to be £4.54 (Dutch value [which is in line with other sources in Europe] converted to GBP due to lack of UK-specific data). Cost of each genetic test, which was used for confirmation after a positive heel-prick screening test result, assumed to be £1,200 (based on prices from Oxford Genetic Laboratories assuming both gene sequencing and multiplex ligation-dependent probe amplification were needed). Treatment and administration costs are based on UK list prices and the latest NHS reference costs (2019/2020).[4]
**Based on the results of a systematic literature review of 21 studies published between 2000 and 2014. Mean age of onset, diagnosis and delay in diagnosis was extracted or calculated. All estimates were weighted by the number of patients and descriptive statistics reported. The weighted mean (standard deviation) ages of onset was 2.5 (0.6) months for SMA Type I, and the weighted mean (standard deviation) age of confirmed SMA genetic diagnosis was 6.3 (2.2) months. The mean delay in diagnosis was 3.6 months.[5]

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