Spinal muscular atrophy: screen at birth, save lives

Author: European Alliance for Newborn Screening in Spinal Muscular Atrophy

Photo by: Christopher Hargoues / AFM-Téléthon

SMA NBS Alliance

The European Alliance for Newborn Screening for SMA demands that by 2025, newborn screening programmes in Europe include a test for spinal muscular atrophy for all newborn children.

In order to advocate for newborn screening for SMA in Europe, SMA Europe founded the SMA NBS Alliance in August 2020 to bring together all stakeholders who share this vision and are willing to work together towards making it a reality.

The overarching objective of the Alliance is to reduce the time it takes for a child born with spinal muscular atrophy to be diagnosed, and to assist patient advocacy groups in their efforts to accelerate the identification of such children, given that early diagnosis and treatment of spinal muscular atrophy leads to better outcomes.

Background

What is SMA?

Spinal muscular atrophy (SMA) is a rare, progressive, neuromuscular disease that leads to immobility and results in a short life expectancy for many children diagnosed with the disease.

Why screen for SMA?

SMA can be treated. There are three disease-modifying treatment options for SMA now approved in Europe with more treatments under development. Most children detected and treated in the first week of life can benefit from normal functional development.

Who is part of the Alliance?

SMA Europe established the European Alliance for SMA Newborn Screening and current members include the European Reference Network EURO-NMD, EURORDIS, TREAT- NMD, EAMDA, University of Groningen, Health-Ecore, Novartis Gene Therapies, Biogen, Roche, Perkin Elmer, ImmunoIVD, and LaCAR MDX Therapeutics.

Founding member

SMA Europe

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Members

European Reference Network for rare or low prevalence complex diseases – Network Neuromuscular Diseases (ERN EURO-NMD)
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LaCAR Life and Care Advanced Research
ImmunoIVD
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ibima Instituto de Investigación Biomédica de Málaga

SMA meets the WHO criteria for adding a new disease to screening programmes

1. SMA is an important health problem

- 5q SMA is a rare, genetic disease with an incidence of 1 in 6,000 to 10,000 live births
- Based on age of onset of symptoms and the maximum motor function achieved, SMA is currently classified into four main types which broadly reflect the severity of the condition
- Without treatment and depending on the severity of the condition, babies may not reach two years of age or their ability to sit, walk and breathe may be significantly impaired. SMA is therefore an important health problem

2. There are accepted treatment options for patients with SMA

- Three disease-modifying treatment options for SMA have now been approved in Europe
- More treatments are under development
- There is growing evidence that earlier treatment leads to greater potential outcomes

3. Facilities for diagnosis and treatment of SMA are available

- There are numerous health care institutions across Europe that provide state-of-the-art care to people living with SMA

4. There is a recognisable latent or early symptomatic stage of SMA

- There is a time window between birth and age of symptom onset. However, even before the first symptoms, damage to the motor neurons may have already occurred
- This “window of opportunity” is often wasted due to the unavailability of newborn screening

5. There is a suitable newborn screening test for SMA

- A reliable blood test is available for use in SMA newborn screening
- The test identifies a homozygous SMN1 exon 7 deletion
- The sensitivity of this test is estimated to be 95% and specificity is nearly 100%. This means that false positives are very unlikely to occur
- It is a simple, inexpensive (approximately 3-5 Euros), automated and high-throughput test

6. SMA newborn screening is acceptable to the population

- Studies demonstrate that SMA newborn screening is acceptable to the general population

7. The natural history of SMA, including its development from latent to diagnosed disease, is adequately understood

- Sufficient information on the natural history of SMA is available
- Subject to its type, SMA inevitably effect children and causes a marked delay or complete halt in the development of neuromuscular function early in life
- Without early diagnosis and treatment, children with SMA may suffer from severe impairment, accumulation of comorbidities or early death

8. There is an agreed policy on whom to treat

- “Treatment” is not limited to disease modifying drugs only but includes best-supportive care including non-pharmacological treatment (e.g., specialised physiotherapy)
- Treatment is a shared decision-making process between the SMA experts and the child´s parents
- The number of SMN2 copies (a paralogous gene to SMN1 which can partially replace its function) on its own is not sufficient to decide on a treatment with disease-modifying drugs

9. The cost of case finding (including diagnosis) by SMA newborn screening is economically balanced in relation to possible expenditure on health care as a whole

- Newborn screening for SMA can change to can be implemented without major costs, through the dried blood spot specimen already taken for newborn screening
- The cost of screening outweighs the cost of illness
- Detecting SMA early and treating promptly may also save money for health care systems, in addition to improving the quality of life of treated children

10. Case finding is a continuing process and not a “once and for all” project

- Once a newborn screening programme for SMA has started in a country, it should be made available for all babies born in that country from that point onwards
- Introducing SMA newborn screening contributes towards a more inclusive health care system

There is no more time to waste for babies born with SMA - newborn screening programmes for SMA in all European countries by 2025!

Read the full Whitepaper:
SMA: Screen at birth, save a life.