Opening a new horizon for children born with spinal muscular atrophy

Author: European Alliance for Newborn Screening in Spinal Muscular Atrophy

SMA NBS Alliance

Early detection of SMA through newborn screening dramatically improves the effectiveness of all available treatments for this disease

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.

What is newborn screening?
Newborn screening (NBS) is the practice of testing all babies in their first days of life for certain disorders that are treatable, but difficult or impossible to detect clinically. This allows for the identification of patients before the first symptoms emerge and to significantly better treatment outcomes when treated early.

Photo by: Daniel Kostan

Why NBS for SMA?

- 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. This is especially critical in early-onset SMA, where motor neuron degeneration starts before birth and escalates quickly. Starting treatment at this time means the child has a chance of a reasonably normal life without developing all disease complications.
- SMA can be added to the current neonatal heel prick test for a reasonable cost. This cost has to be compared to the psychological and societal impact of the death of babies in the first years of their life, and the lifetime cost of caring for a child with a serious disability.
- Various scientific studies around the world show that treating patients before the onset of the first symptoms could preserve the nervous system and prevent the onset of functional disabilities.
- Most children detected and treated in the first week of life can benefit from normal functional development.
- Tests of good quality to detect SMA exist with testing for SMA already available in many parts of the United States and some pilot studies are ongoing in Europe.
- Effective treatments for SMA are available: Early detection and diagnosis may mean the difference between life and death for an infant born with SMA.

European Alliance for Newborn Screening in SMA

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 decrease 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.

Founding member

SMA Europe

Members

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SMA: Test at birth, save a life.

Identifying children living with SMA when pre-symptomatic through NBS allows for treatment to be started on time, preventing motor neuron death and a life with severe disabilities.

Visit www.sma-europe.eu for more information

Also, join the SMA Europe Mailing List for Updates on the Alliance's Activities