Newborn Screening for Spinal Muscular Atrophy (SMA)

In its most severe forms, spinal muscular atrophy (SMA) can progress rapidly. But the early signs can sometimes be subtle, and may even go unseen for weeks or months as the disease progresses.

Early diagnosis of SMA through newborn screening (NBS) can help detect the disease before symptoms appear, potentially transforming the lives of patients and their families. Novartis Gene Therapies recognizes the urgent need to “screen the unseen” and partner with key stakeholders across the globe to ensure that every baby, worldwide, can be screened at birth.

What is newborn screening?

The aim of NBS is to **detect treatable conditions in infants** that are not clinically evident during the newborn period, in order to intervene as early as possible.[1]

This is achieved by a simple blood test **24 to 48 hours** after the baby is born.[1]

Importance of newborn screening for SMA

Diagnosing and treating SMA quickly is crucial to stopping progression of the disease, which robs infants of valuable motor neurons that allow them to walk, sit and even breathe.[2,3,4]

With SMA being one of the leading global genetic causes of infant death, it is crucially important to screen for this condition.[2]

Advances in research and treatment options for SMA

Advances in understanding and treating SMA have encouraged the SMA community to call for a renewed commitment to implement NBS on a global scale. These advances include:

- Development of **life-saving interventions**
- **Early treatment** having a major influence on outcomes
- **Rapid developments** in R&D to identify other treatments[5]
- **Introduction of screening** in the US and other countries globally[6]

Snapshot of newborn screening efforts

Here is an overview of newborn screening programs as of March 2023.

- **North America**: the United States and Canada are screening for SMA. In 2018, SMA was added to the U.S. Recommended Uniformed Screening Panel (RUSP). Currently 48 of the 50 states screen for SMA.[7] In Canada, the majority of provinces now screen for the disease at birth.
- **Europe**: 45% of all newborn babies across Europe are now screened for SMA at birth. Novartis Gene Therapies continues to partner with governments and advocacy groups to increase the number of countries permanently screening.[5, 8]
- **Asia Pacific**: all countries but Myanmar and Laos have national newborn screening programs, however, they do not include SMA. Taiwan and Australia have extensive screening programs for SMA which cover 80% and 44% of newborns respectively.[6]
- **Middle East and North Africa**: newborn screening programs exist but they do not screen for SMA. Somalia and Sudan have no national NBS programs. Little information for newborn screening in Sub-Saharan Africa exists.[9]
- **Latin America**: the majority of countries in Latin America have national and/or regional newborn screening programs.[10]

Collaborating with industry and patient advocacy groups on newborn screenings for SMA

Global

Novartis Gene Therapies has worked with leading rare disease advocacy organizations on matters of policy and patient support. We gladly lend our resources and voice to activities that benefit families impacted by rare disease.

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Europe

In Europe, Novartis partners with SMA Europe, the umbrella organization for European patient organizations for SMA. SMA Europe launched the European Alliance for Newborn Screening in SMA this year, with the goal of including SMA in **all national newborn screening**

programs by 2025. The alliance includes a broad range of organizations, academia and industry.[11]

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References:

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