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 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?

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.\(^2,3\)

With SMA as the leading global genetic cause 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\(^4\)
- Introduction of screening in the US and other countries globally\(^5\)

Snapshot of newborn screening efforts

Here is an overview of newborn screening programs as of December 2020.

- **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 33 of the 50 United States screen for SMA.\(^7\) In Canada, Ontario started screening newborns for SMA in January 2020.\(^8\)

- **Europe**: all countries in Europe have newborn screening programs.\(^9\) The Netherlands, Germany, Poland and Serbia have decided to screen for SMA and implementation planning is underway.\(^10,11,12\) Spain and Belgium have active pilot programs for SMA.\(^10,11,12\)
- **Asia Pacific**: all countries but Myanmar and Laos have national newborn screening programs, though none of these countries screen for SMA. In India, Bangladesh, Cambodia, Indonesia, Pakistan and Vietnam, screening covers less than 5% of newborns.13
- **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.13
- **Latin America**: all but four countries (Dominican Republic, El Salvador, Haiti and Honduras) have national and/or regional newborn screening programs.13

**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.

Learn More

**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 Spinal Muscular Atrophy this year, with the goal of including SMA in **all national newborn screening programmes** by 2025. The alliance includes a broad range of organizations, academia and industry.6

Learn More

**Related Links**

**About Novartis Gene Therapies**

**Understanding Spinal Muscular Atrophy**

**Gene Therapy**

References:

List of links present in page

- https://www.sma-screening-alliance.org/
- http://rarediseases.org/rarediseases/spinal-muscular-atrophy/
- https://www.curesma.org/newborn-screening-for-sma/