



# Newborn Screening in the United States

## Key To Early Diagnosis and Intervention for Neuromuscular Conditions, Including SMA and Pompe Disease

**Newborn screening (NBS)** is a nationwide public health program that identifies newborns with certain serious and life-threatening genetic diseases that can be treated, and for which early treatment may contribute to better outcomes.

The U.S. Secretary of Health and Human Services recommends a list of specific disorders that all states should screen for at birth, collectively called the **Recommended Uniform Screening Panel (RUSP)**. Diseases on the RUSP have been carefully considered through an evidence review process by the Advisory Committee on Heritable Disorders in Newborns and Children, which makes formal recommendations on whether a specific disorder should be added to the RUSP. Each state then looks to the RUSP as a guide to help determine which disorders to include in their individual state NBS programs.

MDA advocates for the inclusion of neuromuscular diseases on the RUSP, and, as a result, two have been added to the list in recent years: **Spinal Muscular Atrophy (SMA)** and **Pompe disease**. We are also working towards addition of other neuromuscular conditions, including **Duchenne Muscular Dystrophy**.

There are currently therapies available for both SMA and Pompe, so it is imperative that babies born with these disorders are identified early to begin receiving treatment as soon as possible. MDA's vast network of **more than 150 MDA Care Centers** sees thousands of patients each year and stands ready to provide treatment to babies identified through newborn screening.

### SMA & Pompe by the numbers

#### SMA

- #1 genetic cause of death for infants
- 1 in 10,000 babies born every year have SMA
- 1 in every 50 people carry the mutation that causes SMA
- 7 states screen babies for SMA
- Added to the national RUSP in 2018

#### Pompe

- Affects 1 in every 138,000 babies born every year
- 16 states screen babies for Pompe
- Added to the national RUSP in 2015



## State Newborn Screening for SMA and Pompe

STATE	SMA	POMPE	STATE	SMA	POMPE
Alabama	X	X	Montana	X	X
Alaska	X	X	Nebraska	X	✓
Arizona	X	X	Nevada	X	X
Arkansas	X	X	New Hampshire	X	X
California	X	✓	New Jersey	X	X
Colorado	X	X	New Mexico	X	X
Connecticut	X	X	New York	✓	✓
Delaware	X	X	North Carolina	X	✓
Florida	X	X	North Dakota	X	X
Georgia	X	X	Ohio	X	✓
Hawaii	X	X	Oklahoma	X	X
Idaho	X	X	Oregon	X	X
Illinois	X	✓	Pennsylvania	✓	✓
Indiana	✓	X	Rhode Island	X	X
Iowa	X	X	South Carolina	X	X
Kansas	X	X	South Dakota	X	X
Kentucky	X	✓	Tennessee	X	✓
Louisiana	X	X	Texas	X	X
Maine	X	X	Utah	✓	X
Maryland	X	X	Vermont	X	X
Massachusetts	X	X	Virginia	X	✓
Michigan	X	✓	Washington	X	X
Minnesota	✓	✓	West Virginia	X	X
Mississippi	X	✓	Wisconsin	X	✓
Missouri	✓	✓	Wyoming	X	X

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