

INTHENews

Why Advocate for Newborn Screening?

This program is crucial for early diagnosis and intervention in some neuromuscular diseases

Newborn screening (NBS) is one of the most impactful public health programs in the United States, especially for the neuromuscular diseases **spinal muscular atrophy** (SMA) and **Pompe disease**. Through NBS, infants born with these diseases can benefit from therapies early in life, improving their health outcomes.

Early diagnosis and treatment for neuromuscular disorders that can be treated is crucial, as the disorders are progressive and, in many cases, fatal. SMA, for example, is the leading genetic cause of death in infants. While only a few years ago there were no disease-modifying therapies for SMA, today there are treatment options, including the first gene therapy ever approved for a neuromuscular condition — approved by the FDA in May — in addition to the first intervention to treat SMA approved by the FDA in 2016. Having two treatment options represents an unprecedented advancement for SMA patients.

How Screening Is Determined

The U.S. Secretary of Health and Human Services recommends a list of disorders that all states should screen infants for at birth, known as the **Recommended Uniform Screening Panel** (RUSP). To be listed on the RUSP, a disease must undergo a rigorous evidence review showing the existence of a reliable confirmatory test, availability of an approved effective treatment, and evidence that early intervention can improve health outcomes and reduce morbidity.

Pompe disease was added to the RUSP in 2015, and SMA was added in 2018. NBS is the key to quickly identifying infants born with these conditions so treatment can be swiftly initiated to realize the full benefits to stave off effects of the disease.

However, there remain challenges to the adoption of NBS for neuromuscular diseases. While the RUSP is a national recommendation, it is ultimately up to each state to determine which disorders they screen for. Currently, 49 states and the District of Columbia screen for at least 31 of the 35 conditions on the RUSP, but only 17 states screen newborns for Pompe disease and eight states screen for SMA.

Advocating for Neuromuscular Diseases

MDA is working to encourage implementation of



screening for neuromuscular conditions through our advocacy efforts and by supporting the bipartisan **Newborn Screening Saves Lives Reauthorization Act of 2019**. This act will reauthorize the federal national NBS program, which expires in September of this year, and provide funds to states so they can update and improve their NBS programs. The bill also authorizes the federal grant program that improves the follow-up process to ensure identified infants are receiving the care they need, and MDA stands ready to provide care and support services for these children and their families with our network of more than 150 Care Centers across the country.

As of July, the U.S. House of Representatives unanimously moved to pass the Newborn Screening Saves Lives Reauthorization Act (HR 2507). We now turn our attention to the Senate.

Although the decision to implement screening for conditions on the RUSP is determined at the state level, the federal government recommendations put pressure on each state to implement these screens, so it is incredibly important that the national NBS program be as robust as possible. Right now, MDA is also working toward the goal of adding screening for **Duchenne muscular dystrophy** (DMD) to the RUSP. Passage of the Newborn Screening Saves Lives Reauthorization Act will help move this forward and secure the overall program for years to come.

Advocate now for the passage of the Newborn Screening Saves Lives Reauthorization Act.