



Newborn Screening Improves Health Outcomes

Newborn screening can change the life of a baby with a neuromuscular disease.

Take spinal muscular atrophy (SMA) as an example. “The difference between getting treatment at six weeks and six months is often the difference between walking or only sitting,” says Diana Castro, MD, a neurologist with special qualifications in Child Neurology and previously an associate professor of Pediatrics and Neurology at University of Texas Southwestern Medical Center. “We understand the natural history of SMA and, though the condition is rare and there will be deterioration, treatment changes the quality of life when it comes to the loss of motor neurons.”

Recommended screening

SMA is one of two neuromuscular diseases included on the Recommended Uniform Screening Panel (RUSP) list, however each state’s public health department determines which conditions are included in its newborn screening panel. All but a few states screen for

SMA. (See [conditions screened by state](#).)

Pompe disease also is included on the RUSP, but currently, about half of states offer screening for Pompe. Screening for this disease is important because infantile-onset Pompe disease can lead to heart failure within the first year of life. Enzyme replacement therapy treatments can help preserve strength and keep heart muscle cells from dying.

SMA and Pompe were only added to the RUSP in the last decade. “You can tell opinions are changing in terms of the importance of screening,” Dr. Castro says. “But it will take time for more to be added.”

The primary barrier to more babies receiving newborn screening is cost. “But a test can be piggybacked on tests already performed for newborn screening,” she says.

Lacking sufficient support staff is another common source of stress. “You sometimes see physicians having to do so much paperwork that they shouldn’t be doing,” she says.

Increasing access to screening

Neurologists, pediatricians, and other health professionals can help families get access to and participate in screenings through continued education. “We must make the public more aware, but it’s also about the pediatrician having one-on-one conversations and explaining what can feel like complex information in a simpler way,” Dr. Castro says. “If they can be told why newborn screening is important and a little bit about these diseases, that would help.”

When it comes to state and federal policy, the best advocacy comes from patients and families, the medical community, and others who are affected by these diseases contacting lawmakers to help them understand what children go through and to explain the difference early treatment makes.

“Showing outcomes is a key,” Dr. Castro says. “For a child to not get the help they need here, this needs to change.”

Visit MDA’s [Action Center](#) to learn how MDA is advocating for access to care and therapies from day one, such as urging lawmakers to support the Newborn Screening Saves Lives Reauthorization Act of 2021, and how you can get involved.