

Newborn Screening's Role in Connecting Children to Expert Care

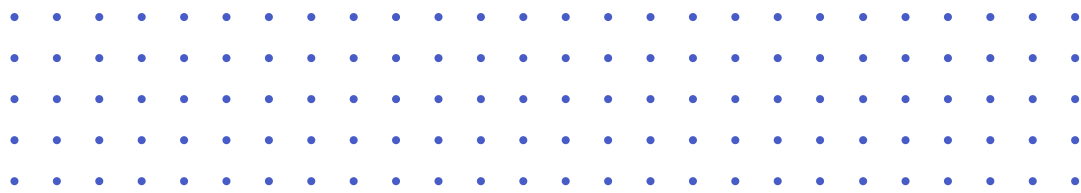
The evolution of newborn screening over the last decade has been good news for neuromuscular diseases (NMDs). Two NMDs have been added to the Recommended Uniform Screening Panel (RUSP) — Pompe disease in 2015 and spinal muscular atrophy (SMA) in 2018 — and Duchenne muscular dystrophy (DMD) is being considered.

While states design their own newborn screening panels, they look to the RUSP for guidance. Currently, 47 states screen for Pompe, and three states have taken the initiative to add DMD to their screening panels, with more planning to do so. Yet, the biggest success has been SMA.

“We saw full implementation of SMA screening across the US as of January last year,” says Bo Hoon Lee, MD, a pediatric neurologist at the University of Rochester. “We’re diagnosing and treating babies presymptomatically, and we get to watch many of these babies reach milestones on typical developmental timelines.”

Newborn screening plays a pivotal role in ensuring that every child can connect with expert clinical care as soon as possible. Dr. Lee notes one encouraging trend she’s seeing: States and neuromuscular providers are working together to ensure prompt, appropriate care for children identified through newborn screening.

“For example, in New York, when SMA screening began, the state newborn screening program designated eight centers across the state to directly receive these referrals. This is a change from the previous pathway, which went to general genetic clinics,” she says. “This network not only streamlined care for SMA-identified babies, which meant that the majority of babies would be seen by a neuromuscular specialist the day following a positive screen, but it connected us as specialists, so we could collaborate on the clinical questions that came up, the longitudinal follow-up, and even provide cross-institutional coverage and planning.”



Lessons learned from this collaboration go beyond SMA. Anticipating a decision on adding DMD to the RUSP, NMD providers are working to establish more robust, evidence-based clinical guidelines for the treatment and management of neuromuscular conditions identified through newborn screening and to build the clinical follow-up pathway.

“I’m excited to see where we’re going to be in five years,” Dr. Lee says. “I think there’s a lot to learn.”

Resources

- MDA’s [Newborn Screening webpage](#)
- [Recommended Uniform Screening Panel](#)
- Searchable list of [conditions screened by state](#)
- MDA’s Grand Rounds Webinar: [Newborn Screening in the US: Working with What We’ve Got](#)