

Updates on Newborn Screening for Neuromuscular Diseases

This year, Ohio became the first and presently the only state to include Duchenne muscular dystrophy (DMD) in its newborn screening program. “It is a huge moment because patients with DMD are occasionally misdiagnosed, which can result in missed opportunities for clinical trials and early treatment options,” says Hoda Z. Abdel-Hamid, MD, FAAN, Associate Professor of Pediatrics, Neurology, and Clinical and Translational Science in the Division of Child Neurology at the University of Pittsburgh.

Currently, spinal muscular atrophy (SMA) and Pompe disease are the only neuromuscular diseases on the federal Recommended Uniform Screening Panel (RUSP), although states ultimately decide which diseases to include in their newborn screening programs. MDA and other organizations have been advocating to add DMD to the RUSP. Advances in therapies for DMD, including a gene therapy approved in June, are making early diagnosis and treatment critical.

Dr. Abdel-Hamid believes other states may follow Ohio’s example for DMD and that limb-girdle muscular dystrophy (LGMD) or myotonic dystrophy (DM) could be the next neuromuscular diseases to break through in a state. “Clinical trials for LGMD geared toward gene therapy is an encouraging area, and there are also trials for natural history studies,” Dr. Abdel-Hamid says. These opportunities may convince states to expand their newborn screening programs.

For families, expanded newborn screening would give them more options sooner that can affect the health of their child and help the families prepare for the future. “It’s possible that we could see many important newborn screening additions for states not far in the future,” she says. “We have to remember it hasn’t been that long in terms of the change with SMA and Pompe disease. If we now have DMD in a state, it’s headed in a positive direction.”

Resources Visit MDA’s [Advocacy](#) website to learn more about MDA’s efforts to [expand newborn screening programs](#) and download a [Newborn Screening Fact Sheet](#).