



The Future of Pediatric Neuromuscular Disease Treatment

It wasn't so long ago that treatment for pediatric neuromuscular diseases was limited. Today, there has never been a better time to be optimistic. In this article, we look toward the future of pediatric neuromuscular disease treatment.

Saunders Bernes, MD, a pediatric neurologist at Phoenix Children's Hospital, says the treatment landscape has changed in a few key ways over the last 5 years. One of the most significant changes is the expansion of newborn screening. Currently, most US states screen for spinal muscular atrophy (SMA) and Pompe disease, which both have approved treatments that reduce or delay symptoms if they are started early. "That's a big evolution in treatment of neuromuscular disease," Dr. Bernes says.

Other developments have enabled researchers to improve clinical trials for neuromuscular diseases. These include standard validated outcome scores for the majority of the pediatric neuromuscular disorders, defined natural histories for many disorders, and the prevalence of

genetic testing. According to Dr. Bernes, clinical trials for neuromuscular diseases are more numerous than ever, and many trials now have built-in open label extensions or expanded access programs

As far as new treatments to expect to see in the future, Dr. Bernes sees gene replacement therapy and gene editing as promising methods. Different types of exon-skipping therapies that will be better dosed and other types of steroid treatments may be on the horizon as well.

Enhancing our knowledge of how new treatments work for pediatric neuromuscular diseases is the fact that almost all children now in multidisciplinary clinics are followed with disease registries. What we call real-world data, much of which is collected through collaborations between MDA and other organizations, can help refine and standardize treatments over a period of time.

In addition, the variability of symptoms within a single neuromuscular disorder has led to new types of treatments. For example, Dr. Bernes points to the differences between people who are symptomatic with SMA and the three FDA-approved treatments for it: nusinersen (Spinraza), onasemnogene abeparvovec (Zolgensma), and risdiplam (Evrysdi). Similarly, many neuromuscular diseases will require more than one type of medical treatment to benefit as many patients as possible.

Increasingly, treatment will be tailored to a patient's specific genetic diagnosis or other characteristics. "The phenotypes of what we're going to look at in each patient over a long period of time will be significantly different than what we're seeing right now," Dr. Bernes says.

