

# A Look at How Rare Disease Care Is Progressing

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Most, if not all, of the neuromuscular diseases under MDA's umbrella are considered rare, meaning they affect fewer than 1 in 2,000 individuals.

What is the biggest challenge in caring for rare diseases? "It's the lack of information, clinical knowledge, prognosis, and treatment on many of them," says Han Phan, MD, a pediatric neurologist and head of research and principal investigator at Rare Disease Research, an independent clinical research site. "Organizations like MDA, patient advocacy groups, and patient registries have worked to create natural histories to try and improve this, closely observing how conditions progress over time with the hope of closing the gap. But still, many rare diseases have little information available to both clinicians and patients and their families."

On the positive side, knowledge of some neuromuscular diseases has expanded in the last five to 10 years, leading to improved care. For example, the Duchenne muscular dystrophy (DMD) disease registry has been critical for defining the disease progression, which has helped improve the standard of care. Spinal muscular atrophy (SMA) and amyotrophic lateral sclerosis (ALS) have also been strong in building a natural history and data and have seen important improvements, to name a few.

Gene therapy may be transforming treatment for rare diseases. The first gene replacement therapy for a neuromuscular disease, onasemnogene abeparvovec (Zolgensma), has been shown to decrease the need for respiratory support and improve motor strength, thereby increasing life expectancy in young children with SMA. And many more therapies are being studied for other diseases.

"Gene therapy is promising because most rare diseases are monogenic — they are controlled by a single gene," Dr. Phan says.

Providers can help families get an accurate diagnosis, which is a crucial first step in getting appropriate treatment for a rare disease. "Time of diagnosis to time of treatment is where time is of the essence for many families," she says.

For rare diseases lacking established natural history data, providers often rely on anecdotal information and focus on treating symptoms. In these cases, Dr. Phan emphasizes that it is especially important to listen to patients and learn from them.

"As providers, we are obligated to learn and provide the best care, and currently, only about 5% of rare diseases have treatments," she says. "Focus and patience will be key as we aim to improve outcomes by advancing innovative therapies for patients with rare diseases."

## MOVR and Rare Diseases

To address the lack of information on rare diseases, MDA established a unified patient registry for neuromuscular diseases (NMDs), the Neuromuscular Observational Research (MOVR) Data Hub. Powered by MDA's network of Care Centers, MOVR aggregates clinical, genetic, and patient-reported data for multiple NMDs to improve health outcomes and accelerate drug development.

Currently, more than 60 active MOVR sites are collecting data on seven NMDs. Researchers are using MOVR data to build predictive algorithms for disease progression, identify discrepancies in care across different demographics, identify eligible participants for clinical trials, and more.

### To learn more about MOVR:

- Visit [mda.org/science/movr](https://mda.org/science/movr).
- Read MOVR publications:
  - MDA MOVR: [Design, Methods, and Initial Observations](#) (*Journal of Neuromuscular Diseases*, 2023).
  - MOVR: [NeuroMuscular ObserVational Research, a unified data hub for neuromuscular diseases](#) (*Genetics in Medicine*, 2019).