

Pediatric Neurology is a specialized field focused on rare disorders, particularly orphan diseases. Orphan diseases are defined as conditions that affect fewer than 200,000 individuals within the United States. Despite their rarity, these diseases impact over 300 million people globally. Of the approximately 7,000 diseases cataloged by the National Institutes of Health's Office of Rare Diseases, a significant proportion are neurological and of genetic origin. Notably, 90% of orphan diseases have severe neurological implications.

Neurological orphan diseases are characterized by their fatality, significant decline in quality of life, and prolonged periods of disability. The diagnosis and management of these rare central nervous system (CNS) disorders are particularly challenging due to limited access to diagnostic genomic sequencing, screening tests, and specialized care. Most neurologic orphan diseases currently lack effective treatments to halt disease progression.

However, the future of diagnosing and managing neurologic orphan diseases holds promise, with advancements in gene-targeting therapies such as CRISPR/Cas9, antisense oligonucleotides (ASO), adeno-associated viruses (AAV), and mammalian target of rapamycin (mTOR) inhibitors offering hope for more effective therapeutic options.

In this context, the Rare Disease Advisory Council (RDAC) can serve as a valuable platform for advocating on behalf of patients with these conditions, helping to bridge gaps in care and support the advancement of treatment options.