Pediatric neuromuscular disorders: Advances in treatment and care.

Martina Ricci*

Department of Pediatrics, Kaohsiung Medical University, Taiwan

Introduction

Pediatric neuromuscular disorders encompass a wide range of conditions that affect the peripheral nervous system, muscles, or the neuromuscular junction in children. These disorders can have profound impacts on motor function, respiratory health, and overall quality of life. Recent advances in treatment and care have significantly improved outcomes for many children with these conditions, providing hope where there was once little [1].

Neuromuscular disorders in children can be classified into several categories, including muscular dystrophies, spinal muscular atrophies, congenital myopathies, and peripheral neuropathies. Each of these conditions has distinct genetic and clinical characteristics: Muscular Dystrophies: This group includes Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD), which are caused by mutations in the dystrophin gene. These conditions lead to progressive muscle weakness and degeneration [2].

Spinal Muscular Atrophy (SMA): SMA is a genetic disorder characterized by the loss of motor neurons in the spinal cord, leading to muscle atrophy and weakness. The severity of SMA varies, with Type 1 being the most severe and Type 4 the least. Congenital Myopathies: These are a heterogeneous group of disorders present at birth that cause muscle weakness and structural abnormalities in muscle fibers. Examples include central core disease and nemaline myopathy [3].

Peripheral Neuropathies: Conditions like Charcot-Marie-Tooth disease (CMT) fall into this category. They involve damage to the peripheral nerves, leading to muscle weakness, atrophy, and sensory loss. One of the most promising areas of advancement in treating pediatric neuromuscular disorders is genetic therapy. These therapies aim to address the root cause of the disorder by correcting or compensating for the genetic mutations involved [4].

Gene Replacement Therapy: This approach involves delivering a functional copy of a gene to compensate for the defective one. For example, onasemnogene abeparvovec (Zolgensma) is a gene therapy approved for treating SMA. It delivers a copy of the SMN1 gene, which is missing or defective in SMA patients, via an adeno-associated virus (AAV) vector [5].

Exon Skipping: This technique uses antisense oligonucleotides (ASOs) to skip over faulty exons during the transcription of genes. Eteplirsen (Exondys 51) is an ASO used in DMD to skip exon 51, allowing the production of a partially functional dystrophin protein. Gene Editing: CRISPR-Cas9 and other gene editing technologies hold promise for directly correcting genetic mutations. Although still in the experimental stage, these technologies have shown potential in preclinical studies for conditions like DMD [6].

Pharmacological treatments have also seen significant advancements. These therapies can help manage symptoms and improve the quality of life for children with neuromuscular disorders. Nusinersen (Spinraza): This drug is an ASO that increases the production of the survival motor neuron (SMN) protein by modifying the splicing of SMN2 pre-mRNA. It has shown significant benefits in improving motor function in children with SMA [7].

While genetic and pharmacological treatments are crucial, comprehensive supportive care remains the cornerstone of managing pediatric neuromuscular disorders. Advances in supportive care have greatly enhanced the quality of life and lifespan for many affected children. Respiratory Support: Non-invasive ventilation (NIV) and cough assist devices have become standard in managing respiratory complications. These interventions help maintain adequate ventilation and reduce the risk of respiratory infections [8].

Nutritional Support: Proper nutrition is vital for children with neuromuscular disorders, as they are at risk of malnutrition due to swallowing difficulties and increased metabolic demands. Gastrostomy tubes and nutritional supplements are commonly used to ensure adequate intake. Orthopedic Interventions: Scoliosis is a common complication in many neuromuscular disorders. Early detection and intervention, including bracing and surgical correction, can prevent severe deformities and improve respiratory function [9].

Biomarker Development: Biomarkers can help monitor disease progression and response to treatment. For example, blood levels of creatine kinase (CK) are used to monitor muscle damage in DMD. Neuroimaging: Advanced imaging techniques such as MRI and ultrasound provide detailed views of muscle and nerve structures, aiding in the diagnosis and monitoring of neuromuscular disorders [10].

Conclusion

The landscape of pediatric neuromuscular disorders is rapidly evolving, with significant advances in genetic therapies, pharmacological treatments, supportive care, diagnostic tools,

***Correspondence to:** Martina Ricci, Department of Pediatrics, Kaohsiung Medical University, Taiwan. E-mail: martina@kmu.TW.in

***Received:** 24-Apr-2024, Manuscript No. JNNR-24-137841; **Editor assigned:** 25-Apr-2024, Pre QC No. JNNR-24-137841(PQ); **Reviewed:** 09-May-2024, QC No. JNNR-24-137841; **Revised:** 13-May-2024, Manuscript No. JNNR-24-137841(R); **Published:** 20-May-2024, DOI: 10.35841/aajnnr-9.3.202*

***Citation:** Ricci M. Pediatric neuromuscular disorders: Advances in treatment and care. J Neurol Neurorehab Res.2024;9(3):202*

and psychosocial support. These advancements offer new hope and improved outcomes for children affected by these challenging conditions. Continued research and innovation will undoubtedly lead to even more effective treatments and a better quality of life for these young patients and their families.

References

1. Waldrop MA, Flanigan KM. Update in Duchenne and Becker muscular dystrophy. Curr Opin Neurol. 2019;32(5):722-7.
2. Markati T, Oskoui M, Farrar MA, et al. Emerging therapies for Duchenne muscular dystrophy. Lancet Neurol. 2022;21(9):814-29.
3. Chikkannaiah M, Reyes I. New diagnostic and therapeutic modalities in neuromuscular disorders in children. Curr Probl Pediatr Adolesc Health Care. 2021;51(7):101033.
4. Hor KN, Mah ML, Johnston P, et al. Advances in the diagnosis and management of cardiomyopathy in Duchenne muscular dystrophy. Neuromuscul Disord. 2018;28(9):711-6.
5. Bourke J, Turner C, Bradlow W, et al. Cardiac care of children with dystrophinopathy and females carrying DMD-gene variations. Open Heart. 2022;9(2):001977.
6. Leon-Astudillo C, Okorie CU, McCown MY, et al. ATS core curriculum 2022. pediatric pulmonary Medicine: updates in pediatric neuromuscular disease. Pediatr Pulmonol. 2023 ;58(7):1866-74.
7. Sadat R, Emrick L. Genetic testing and counseling and child neurology. Neurol Clin. 2021;39(3):705-17.
8. Varnier GC, Pilkington CA, Wedderburn LR. Juvenile dermatomyositis: novel treatment approaches and outcomes. Curr Opin Rheumatol. 2018;30(6):650-4.
9. Farrar MA, Calotes-Castillo L, De Silva R, et al. Gene therapy-based strategies for spinal muscular atrophy—an Asia-Pacific perspective. Mol Cell Pediatr. 2023;10(1):17.
10. Mercuri E. Spinal muscular atrophy: from rags to riches. Neuromuscul Disord. 2021;31(10):998-1003.

Citation: *Ricci M. Pediatric neuromuscular disorders: Advances in treatment and care. J Neurol Neurorehab Res.2024;9(3):202*