Duchenne muscular dystrophy: Exploring the pathogenesis and therapeutic strategies.

Stanley Beers*

Department of Physiology, University of Oxford, UK

Introduction

Duchenne Muscular Dystrophy (DMD) is one of the most severe and common forms of muscular dystrophy, a group of genetic disorders characterized by progressive muscle degeneration and weakness. DMD primarily affects boys and is caused by mutations in the DMD gene, which encodes the protein dystrophin. This essential protein helps maintain the integrity of muscle fibers, and its absence leads to the severe symptoms associated with the disease [1].

The DMD gene is located on the X chromosome, which is why the disorder predominantly affects males. Females, who have two X chromosomes, can be carriers of the mutation but typically do not exhibit severe symptoms because they have a second, normally functioning copy of the gene. When dystrophin is absent or severely reduced due to mutations, muscle fibers become damaged and weakened over time. This damage results in chronic inflammation, fibrosis (formation of excess fibrous connective tissue), and eventual replacement of muscle tissue with fat and connective tissue [2].

The onset of DMD symptoms usually occurs in early childhood, typically between the ages of 2 and 5. The progression of the disease is predictable, with muscle weakness and degeneration starting in the proximal muscles (those closer to the center of the body) and later affecting distal muscles (those farther from the center). Common early signs and symptoms include: Delayed Motor Milestones: Children with DMD often exhibit delayed walking and difficulty running, jumping, and climbing stairs [3].

Gower's Sign: A characteristic way of rising from the floor, where the child uses their hands to push on their thighs to stand up due to weak hip and thigh muscles. Enlarged Calves (Pseudohypertrophy): Despite appearing larger, the calf muscles are weak and contain increased amounts of fat and connective tissue. Frequent Falls: Due to muscle weakness and lack of coordination [4].

Diagnosing DMD involves a combination of clinical evaluation, family history, and specialized tests. Key diagnostic procedures include: Creatine Kinase (CK) Test: Elevated levels of CK, an enzyme released from damaged muscle, can indicate muscle disease [5].

Genetic Testing: Identifying mutations in the DMD gene confirms the diagnosis. Muscle Biopsy: Occasionally performed to examine the muscle tissue for the absence of dystrophin protein. Electromyography (EMG): To assess the electrical activity of muscles and rule out other neuromuscular disorders [6].

DMD is caused by mutations in the DMD gene, which provides instructions for making dystrophin. Dystrophin is crucial for maintaining the stability and structure of muscle cell membranes during contraction and relaxation. The gene's mutations can be deletions, duplications, or point mutations, all leading to the absence or insufficient production of functional dystrophin [7].

While there is currently no cure for DMD, treatment focuses on managing symptoms, improving quality of life, and prolonging survival. A multidisciplinary approach is essential, involving neurologists, cardiologists, pulmonologists, physical therapists, and other specialists. Key treatment strategies include: Corticosteroids: Drugs like prednisone and deflazacort can slow muscle degeneration and improve strength and function [8].

Orthopedic Interventions: Surgeries to correct skeletal deformities and improve function. Assistive Devices: Use of wheelchairs, braces, and other aids to enhance mobility and independence. Research and Emerging Therapies Research into DMD is vigorous, with many promising therapies under investigation. Some of the most notable areas of research and emerging treatments include: Gene Therapy: Techniques aimed at delivering a functional copy of the DMD gene to muscle cells [9].

Stem Cell Therapy: Investigating the potential of stem cells to regenerate damaged muscle tissue. CRISPR/Cas9 Gene Editing: Research is ongoing into using this advanced technology to correct mutations in the DMD gene at the DNA level. Utrophin Modulation: Utrophin is a protein similar to dystrophin. Drugs that increase utrophin production are being explored as potential treatments. Anti-inflammatory and Anti-fibrotic Agents: Aimed at reducing inflammation and fibrosis in muscle tissue [10].

Conclusion

Duchenne Muscular Dystrophy is a devastating genetic disorder that profoundly impacts the lives of patients and their families. While significant advances have been made in understanding the disease and developing treatments that can

***Correspondence to:** Stanley Beers, Department of Physiology, University of Oxford, UK. E-mail: sbeers@uox.uk

***Received:** 25-Dec-2023, Manuscript No. JNNR-24-137395; **Editor assigned:** 27-Dec-2023, Pre QC No. JNNR-24-137395(PQ); **Reviewed:** 10-Jan-2024, QC No. JNNR-24-137395; **Revised:** 15-Jan-2024, Manuscript No. JNNR-24-137395(R); **Published:** 22-Jan-2024, DOI: 10.35841/aajnnr-9.1.182*

***Citation:** Beers S. Duchenne muscular dystrophy: Exploring the pathogenesis and therapeutic strategies. J Neurol Neurorehab Res.2024;9(1):182*

slow its progression, much work remains to be done. Ongoing research holds the promise of more effective therapies and, ultimately, a cure. In the meantime, comprehensive, multidisciplinary care and robust support systems can significantly improve the quality of life for those affected by DMD.

References

1. Sun C, Shen L, Zhang Z, et al. Therapeutic strategies for Duchenne muscular dystrophy: an update. Genes (Basel). 2020;11(8):837.
2. Salmaninejad A, Jafari Abarghan Y, Bozorg Qomi S, et al. Common therapeutic advances for Duchenne muscular dystrophy (DMD). Int J Neurosci. 2021;131(4):370-89.
3. Mbakam CH, Lamothe G, Tremblay G, et al. CRISPR-Cas9 gene therapy for Duchenne muscular dystrophy. Neurotherapeutics. 2022;19(3):931-41.
4. Bez Batti Angulski A, Hosny N, Cohen H, et al. Duchenne muscular dystrophy: Disease mechanism and therapeutic strategies. Front Physiol. 2023;14:1183101.
5. Takeda SI, Clemens PR, Hoffman EP. Exon-skipping in Duchenne muscular dystrophy. J Neuromuscul Dis. 2021;8(2):343-58.
6. Min YL, Bassel-Duby R, Olson EN. CRISPR correction of Duchenne muscular dystrophy. Annu Rev Med. 2019;70:239-55.
7. Osorio AN, Cantillo JM, Salas AC, et al. Consensus on the diagnosis, treatment and follow-up of patients with Duchenne muscular dystrophy. Neurologia (Engl Ed). 2019;34(7):469-81.
8. Crisafulli S, Sultana J, Fontana A, et al. Global epidemiology of Duchenne muscular dystrophy: an updated systematic review and meta-analysis. Orphanet J Rare Dis. 2020;15:1-20.
9. Erkut E, Yokota T. CRISPR therapeutics for Duchenne muscular dystrophy. Int J Mol Sci. 2022;23(3):1832.
10. Sun C, Serra C, Lee G, et al. Stem cell-based therapies for Duchenne muscular dystrophy. Exp Neurol. 2020;323:113086.

Citation: *Beers S. Duchenne muscular dystrophy: Exploring the pathogenesis and therapeutic strategies. J Neurol Neurorehab Res.2024;9(1):182*