

---

# Duchenne Muscular Dystrophy

## Public Education

---

**Correspondence:** Exon Publications, Brisbane, Australia; Email: [books@exonpublications.com](mailto:books@exonpublications.com)

**Cite as:** Duchenne Muscular Dystrophy: Public Education. Brisbane (AU): Exon Publications; 2024. Published on 04 Oct 2024.

DOI: <https://doi.org/10.36255/duchenne-muscular-dystrophy-public-education>

**Copyright:** Exon Publications

**License:** Creative Commons Attribution-NonCommercial-NoDerivs 4.0 (CC BY-NC-ND 4.0)  
<https://creativecommons.org/licenses/by-nc-nd/4.0/>

---

## Abstract

Duchenne Muscular Dystrophy (DMD) is a severe genetic disorder that affects the muscles, causing them to weaken over time. This condition primarily impacts boys and leads to progressive physical disability, requiring comprehensive care and management. This guide provides an accessible yet detailed explanation of DMD, addressing its causes, inheritance patterns, symptoms, diagnosis, treatment, and daily management. The content is designed to serve as a valuable resource for patients, families, and caregivers seeking clarity on this condition. Understanding the role of the dystrophin gene and exploring treatment options,

including emerging therapies, is critical to managing DMD and improving the quality of life for those affected.

## Introduction

Duchenne Muscular Dystrophy (DMD) is a rare but serious genetic condition that primarily affects boys, causing the gradual weakening of muscles throughout the body. It is caused by mutations in the dystrophin gene, which is responsible for producing a protein essential for muscle health and stability. Without dystrophin, muscles become damaged and eventually lose their function. This condition requires a lifetime of care, involving physical therapy, medications, and sometimes surgical interventions to manage symptoms and complications. By raising awareness and understanding about DMD, families can better navigate the challenges it presents and access the most effective treatments available (1-3).

## What is Duchenne Muscular Dystrophy?

Duchenne Muscular Dystrophy is a progressive muscle-wasting disease that primarily affects children. It occurs when the body cannot produce enough dystrophin, a protein crucial for protecting muscle fibers during movement. The absence or deficiency of dystrophin causes the muscles to weaken and deteriorate over time. This condition is part of a group of disorders known as muscular dystrophies, which all involve muscle weakness and degeneration but vary in severity and age of onset. DMD is considered the most common and severe form of muscular dystrophy, significantly impacting mobility, respiratory function, and heart health as the disease progresses. The early signs of DMD typically appear between the ages of two

and five years, with children experiencing difficulty running, jumping, or climbing stairs.

## Epidemiology of Duchenne Muscular Dystrophy

Duchenne Muscular Dystrophy affects approximately one in every 3,500 to 5,000 live male births worldwide. While the condition primarily impacts boys, rare cases have been reported in girls due to specific genetic circumstances. DMD occurs in all ethnic groups and geographical regions, but the prevalence may vary slightly across populations. The genetic mutation responsible for DMD is carried on the X chromosome, which explains why males are predominantly affected, as they have only one X chromosome. Females, with two X chromosomes, are typically carriers of the condition, meaning they can pass the mutated gene to their offspring without exhibiting symptoms themselves.

## Types of Duchenne Muscular Dystrophy

While Duchenne Muscular Dystrophy is generally considered one type of muscular dystrophy, it is distinct from other forms like Becker Muscular Dystrophy, which is caused by similar mutations in the dystrophin gene but results in a milder course of disease. DMD itself does not have subtypes, but its progression can vary slightly between individuals based on the nature of the genetic mutation and other factors. Becker Muscular Dystrophy is often considered a related condition rather than a subtype of DMD, as it involves the partial production of dystrophin, leading to slower muscle degeneration and a later onset of symptoms.

# Genetics and Inheritance of Duchenne Muscular Dystrophy

Duchenne Muscular Dystrophy is caused by mutations in the DMD gene, which is one of the largest genes in the human body. This gene provides instructions for making dystrophin, a protein essential for maintaining the structural integrity of muscle cells. The mutation in the DMD gene either prevents the production of dystrophin or leads to the creation of a non-functional version of the protein. DMD is inherited in an X-linked recessive pattern. Since males have only one X chromosome, a single mutation in the DMD gene is enough to cause the disease. Females, with two X chromosomes, are usually carriers because they have one functional copy of the gene that compensates for the defective one. However, some female carriers may experience mild symptoms due to skewed X-chromosome inactivation.

# Risk Factors and Causes of Duchenne Muscular Dystrophy

The primary cause of Duchenne Muscular Dystrophy is a mutation in the DMD gene located on the X chromosome. This mutation is usually inherited from a parent who is a carrier of the defective gene, but in some cases, it may arise spontaneously without a family history. The most significant risk factor for DMD is being male, as males are more susceptible to X-linked genetic conditions due to their single X chromosome. A family history of DMD increases the likelihood of the condition occurring in offspring, and genetic testing can help identify carriers of the mutation. Environmental or lifestyle factors do not play a role in the development of DMD, as it is purely a genetic disorder.

# Symptoms of Duchenne Muscular Dystrophy

The symptoms of Duchenne Muscular Dystrophy typically appear in early childhood, often between the ages of two and five. Early signs include delays in reaching motor milestones such as sitting, standing, or walking. Children with DMD may have difficulty running, jumping, or climbing stairs and may exhibit a characteristic waddling gait. Other common symptoms include frequent falls, enlarged calf muscles due to fatty tissue replacing muscle fibers, and difficulty getting up from a sitting or lying position. As the disease progresses, muscle weakness spreads to other areas of the body, affecting the arms, respiratory muscles, and heart. Eventually, individuals with DMD may lose the ability to walk and require the use of a wheelchair. Respiratory complications and heart issues become more prominent in the later stages of the disease.

# Diagnosis of Duchenne Muscular Dystrophy

Diagnosing Duchenne Muscular Dystrophy involves a combination of clinical evaluations, laboratory tests, and genetic analysis. A doctor may suspect DMD if a child shows signs of delayed motor development, muscle weakness, or other characteristic symptoms. Blood tests measuring levels of creatine kinase (CK), an enzyme that leaks into the bloodstream when muscle cells are damaged, can indicate muscle degeneration. Genetic testing is the definitive method for diagnosing DMD, as it can identify mutations in the DMD gene. In some cases, a

muscle biopsy may be performed to confirm the absence of dystrophin in muscle tissue. Early diagnosis is critical for initiating appropriate care and treatment to manage symptoms and slow disease progression.

## Treatment and Management of Duchenne Muscular Dystrophy

There is no cure for Duchenne Muscular Dystrophy, but treatments aim to manage symptoms, slow disease progression, and improve quality of life. Corticosteroids, such as prednisone and deflazacort (Emflaza), are commonly prescribed to preserve muscle strength and delay the loss of mobility. Physical therapy and regular exercise are essential for maintaining muscle function and preventing contractures, which are stiffening of the joints due to muscle shortening. Assistive devices such as braces, walkers, and wheelchairs can help maintain mobility as the disease progresses. Respiratory support, including non-invasive ventilation or tracheostomy, may be needed as the respiratory muscles weaken. Cardiac care is crucial, as DMD can lead to cardiomyopathy, and medications such as ACE inhibitors or beta-blockers are often prescribed to manage heart complications. Gene therapy and exon-skipping drugs, such as eteplirsen (Exondys 51), offer promising new approaches by targeting specific genetic mutations to restore some dystrophin production. These treatments are still evolving but represent significant advances in the management of DMD.

# Prognosis of Duchenne Muscular Dystrophy

The prognosis for individuals with Duchenne Muscular Dystrophy has improved over the years due to advances in medical care and supportive therapies. However, DMD remains a life-limiting condition, with most individuals experiencing significant physical disability by their teenage years. In the past, life expectancy for people with DMD was often limited to the late teens or early twenties. With modern medical interventions, including improved respiratory and cardiac care, individuals with DMD can now live into their thirties or even forties in some cases. Early diagnosis, proactive management, and access to specialized care can significantly enhance the quality of life and extend life expectancy for those with DMD.

# Living with Duchenne Muscular Dystrophy

Living with Duchenne Muscular Dystrophy requires a multi-faceted approach to care that addresses physical, emotional, and social needs. Families play a central role in managing the condition, often working closely with a team of healthcare professionals, including neurologists, physical therapists, cardiologists, and respiratory specialists. Daily routines often include physical therapy exercises to maintain muscle flexibility and strength, as well as the use of assistive devices to support mobility and independence. Emotional support is equally important, as individuals with DMD and their families may face significant psychological challenges. Support groups and counseling services can provide valuable resources and a sense of community for those affected by the disease. Education

and advocacy are essential for ensuring that individuals with DMD have access to appropriate care, accommodations, and opportunities throughout their lives.

## Conclusion

Duchenne Muscular Dystrophy is a challenging condition that requires a comprehensive understanding of its genetic basis, symptoms, and management strategies. While there is currently no cure, advances in medical research and treatment offer hope for improving outcomes and quality of life for individuals with DMD. Early diagnosis, proactive care, and access to emerging therapies are critical for managing the disease effectively.

## References

1. Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. *Lancet Neurol.* 2018 Mar;17(3):251-267. [https://doi.org/10.1016/S1474-4422\(18\)30024-3](https://doi.org/10.1016/S1474-4422(18)30024-3)
2. Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management. *Lancet Neurol.* 2018 Apr;17(4):347-361. [https://doi.org/10.1016/S1474-4422\(18\)30025-5](https://doi.org/10.1016/S1474-4422(18)30025-5)
3. Mendell JR, Sahenk Z, Lehman K, et al. Assessment of Systemic Delivery of rAAVrh74.MHCK7.micro-dystrophin in Children With Duchenne Muscular Dystrophy: A Nonrandomized Controlled Trial. *JAMA Neurol.* 2020 Sep 1;77(9):1122-1131. <https://doi.org/10.1001/jamaneurol.2020.1484>



## Notice to the User

This article was written by professional medical writers for the general public based on peer-reviewed articles indexed in PubMed and peer-reviewed for scientific accuracy by independent experts. It is intended solely for informational purposes and is not to be considered medical advice. The views and opinions expressed in this article are believed to be accurate at the time of publication, but the publisher, editors, or authors cannot be held responsible or liable for any errors, omissions, or consequences arising from the use of the information contained in this article. The publisher makes no warranties, implicit or explicit, regarding the contents of this article or its use.