

PHYSIO IN DUCHENNE MUSCULAR DYSTROPHY (DMD)

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ABSTRACT:

Duchenne muscular dystrophy is the most common and severe form of muscular dystrophy and is caused by mutations in the dystrophin gene. Dystrophin, together with several other protein components, is part of a complex known as the dystrophin glycoprotein complex (DGC). The DGC plays an essential role in maintaining the structural integrity of the muscle cell membrane by providing a link between the extracellular matrix and the cytoskeleton. The absence of functional dystrophin and the consequent loss of the DGC ultimately lead to chronic muscle damage resulting in progressive muscle weakness seen in DMD patients. There are currently no long-term effective treatments available and DMD patients usually die from respiratory or cardiac muscle failure in their 20s.

KEYWORDS: *Duchenne muscular dystrophy, dystrophin glycoprotein complex, allelic, dystrophin gene.*

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INTRODUCTION:

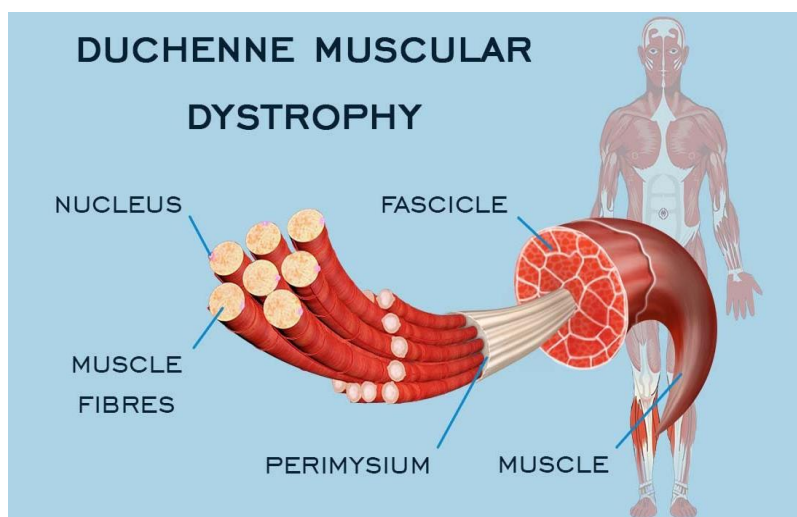
Duchenne and Becker muscular dystrophies are X-linked allelic disorders which are caused by mutations in the DMD gene. It is inherited in an X-linked recessive pattern. X-linked means that the gene for the condition is located on the X-chromosome, one of the sex chromosomes.



In males (who have only one X chromosome), one altered copy of the gene is enough to cause the condition. X-linked recessive conditions affect males much more frequently than females. Females, who have one altered gene, are called carriers. While, most female carriers have no signs or symptoms of the condition, in rare cases, female carriers may experience some mild signs or symptoms. Sometimes a male child is the first person in a family with the condition. In this case, the gene alteration may have been inherited from the mother, or the alteration may have occurred by chance for the first time in the child.

DEFINITION:

Muscular dystrophy (MD) is a Degenerative muscle disease causing progressive weakness, loss of ambulation usually by age 12, and death from respiratory and cardiac failure in the second decade of life.

**EPIDEMIOLOGY:**

- X-linked recessive inheritance
- Most common muscular dystrophy in children

INCIDENCE:

1 in 3500 male new-borns; about 400 to 600 new cases every year.

ONSET OF SYMPTOMS:

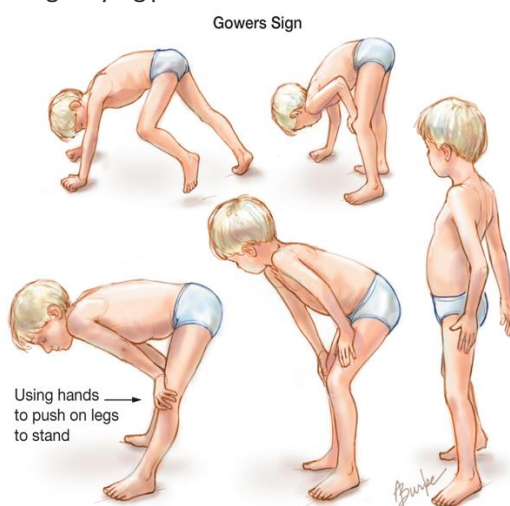
- 3 to 5 years (Duchenne)
- 5 to 15 years or later (Becker)

CAUSES:

- Caused by the absence or deficiency of dystrophin, a subsarcolemmal cytoskeletal protein essential for the histologic integrity and membrane function of skeletal muscle
- Mutations in the dystrophin gene (Xp21 band)
- About one third of cases caused by de novo gene mutation while the rest are inherited in an X-linked recessive pattern

CLINICAL FEATURES:

- Muscle weakness is the principal symptom of DMD. It can begin as early as age 2 or 3, first affecting the proximal muscles (those close to the core of the body) and later affecting the distal limb muscles (those close to the extremities).
- Difficulty getting up from a sitting or lying position



(Gower's sign)

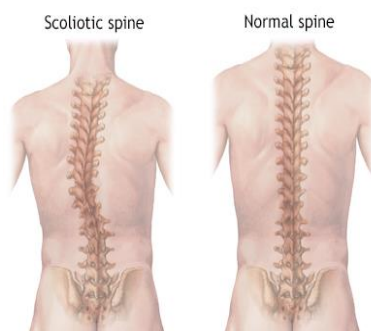
- Delayed walking, toe walking, frequent falls, inability to jump or run, difficulty keeping up with peers
- Waddling gait / Myopathic Gait

Myopathic Gait

- Calf muscle pseudohypertrophy
- Loss of ambulation usually occurs by age 12 in Duchenne MD and after age 15 in BMD.

Other signs include:

- Cardiomyopathy
- Respiratory failure in the late stages
- Scoliosis



- Gastric hypomotility
- Central nervous system manifestations in about one third:
 - delayed speech
 - decreased verbal ability
 - impaired intellectual function (nonprogressive)

Characteristic Features of Duchenne Muscular Dystrophy:

- Enlarged calf muscles
- Shrinking of thigh muscles
- Progressive (worsening) muscle weakness
- Intellectual impairment

DIAGNOSTIC EVALUATION:

- Duchenne muscular dystrophy (DMD) is diagnosed in young boys based on clinical examination, signs and symptoms, family history, and confirmed by the results of genetic testing.
- A muscle biopsy may be done to remove a small piece of muscle for examination under a microscope.
- Blood tests looking for increased levels of certain special proteins called muscle enzymes are used to check for muscle damage.

What are DMD “carriers”?

DMD carriers are females who have a normal dystrophin gene on one X chromosome and an abnormal dystrophin gene on the other X chromosome. Most carriers of DMD do not themselves have signs and symptoms of the disease, but a minority do. Symptoms can range from mild skeletal muscle weakness or cardiac involvement to severe weakness or cardiac effects and can begin in childhood or adulthood

TREATMENT:

- There is no known cure for Duchenne muscular dystrophy (DMD).
- Treatment is focused on managing the symptoms of DMD and related complications caused by severe progressive muscle weakness and loss.
- Medications (such as steroids) may improve the strength and function of muscles.
- Additional medications are available for people with DMD with a specific DNA variant.
- These can help improve muscle strength and function. An enlarged, weakened heart (dilated cardiomyopathy) may be treated with medications, but in severe cases, a heart transplant may be necessary.
- Assistive devices for breathing difficulties may be needed, especially at night and as the disease progresses.
- **Deflazacort** for the treatment of Duchenne Muscular Dystrophy in patients 5 years of age and older.
- **Eteplirsen** for the treatment of Duchenne muscular dystrophy (DMD) in patients who have a confirmed mutation of the DMD gene
- **Golodirsen** for the treatment of Duchenne muscular dystrophy (DMD) in patients who have a confirmed mutation of the DMD gene

ROLE OF PHYSIOTHERAPIST:

Physiotherapy is essential to the management of Duchenne's. It is important to monitor the physical symptoms of the condition and physiotherapy can help keep the child active for as long as possible. Physiotherapists will work with the parents and carers and provide them with information and manual skills that will be helpful for the child.

- **Contractures** are one of the major side effects that a physiotherapist will address. They will do these through a stretching routine, which can also be taught to the parents.
- Physiotherapists will also be responsible for advising the parents on any **orthoses**
- the physiotherapist will be involved in helping **keep the child active**.
- Physiotherapists will monitor the child's **posture** in sitting, lying and standing. They can inform the parents of ways to help the child sit, stand and lie in optimal positions using pillows or splints.
- A sleep system and night splints may be recommended for night time to help maintain the child's posture over a long period of time.

Due to the wasting of the muscle caused by the lack of dystrophin, DMD patients will struggle with many everyday activities. Physiotherapists can help with the management of presenting neuromusculoskeletal problems. They can help slow the regression of range of motion, muscle strength, daily function, work to improve gait pattern and posture/alignment. Physiotherapy can also address the pain that the patient may be experiencing. As the patient's walking and standing abilities decline the physiotherapist may choose to implement a standing program.

CONCLUSIONS:

Duchenne muscular dystrophy (DMD) affects the muscles, leading to muscle wasting that gets worse over time. DMD occurs primarily in males, though in rare cases may affect females. The symptoms of DMD include progressive weakness and loss (atrophy) of both skeletal and heart muscle. Early signs may include delayed ability to sit, stand, or walk and difficulties learning to speak. Muscle weakness is usually noticeable in early childhood. Most children with DMD use a wheelchair by their early teens. Heart and breathing problems also begin in the teen years and lead to serious, life-threatening complications.

REFERENCES:

1. Ryder, S. *et al.* The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: An evidence review. *Orphanet Journal of Rare Diseases* (2017). doi:10.1186/s13023-017-0631-3
2. OGATA, H., ISHIKAWA, Y., ISHIKAWA, Y. and MINAMI, R., 2009. Beneficial effects of beta-blockers and angiotensin-converting enzyme inhibitors in Duchenne muscular dystrophy. *Journal of Cardiology.* , vol. 53, no. 1, pp. 72-78.
3. ORPHANET., 2007. Duchenne and Becker Muscular Dystrophy [online]. [viewed 6 October 2014]. Available from: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=262
4. Moat, S. J., Bradley, D. M., Salmon, R., Clarke, A. & Hartley, L. Newborn bloodspot screening for Duchenne Muscular Dystrophy: 21 years' experience in Wales (UK). *Eur. J. Hum. Genet.* (2013). doi:10.1038/ejhg.2012.301
5. Romitti, P. A. *et al.* Prevalence of Duchenne and Becker Muscular Dystrophies in the United. *Pediatrics* (2015). doi:10.1542/peds.2014-2044
6. Venugopal V, Pavlakis S. Duchenne Muscular Dystrophy. In: StatPearls. StatPearls Publishing, Treasure Island (FL); 2020. PMID: 29493971.
7. Ropars, J *et al.*, (2016) "Muscle activation during gait in children with Duchenne Muscular Dystrophy" vol-11: No :9 .
8. <https://www.sciencedirect.com/topics/medicine-and-dentistry/duchenne-muscular-dystrophy>.
9. https://www.physio-pedia.com/Duchenne_Muscular_Dystrophy