

DUCHENNE

MUSCULAR DYSTROPHY (DMD)

FAMILY FACTSHEET

ABOUT DUCHENNE MUSCULAR DYSTROPHY

1 WHAT IS DUCHENNE MUSCULAR DYSTROPHY?

- Duchenne muscular dystrophy (DMD) is a rare and severe form of muscular dystrophy primarily affecting males.
- It is caused by a mutation in the dystrophin gene located on the X chromosome. Dystrophin is important for muscle cell stability.
- Muscles to become weaker over time until it affects the whole body.
- One in 5,000 boys are born with DMD worldwide.
- The DMD gene mutation can be passed from the mother, or can happen spontaneously.
- A woman with a genetic change in one of her two copies is said to be 'a carrier'.
- DMD is a progressive disease. First, walking becomes difficult. Then, other motor functions follow until it ultimately affects the ability to breathe as well as the function of the heart.

2 WHAT ARE EARLY SIGNS OF DUCHENNE MD?

- Signs of DMD usually are recognized when kids are very young. However, diagnosis often takes place at the age of 4 or even later.
- Symptoms include muscles getting weaker over time, trouble walking, falling down a lot, and being slower to reach functional milestones like crawling or walking.
- The missing protein also has a function in the brain, so learning- and behavior issues can also be part of the disease.
- The diagnosis typically involves a series of clinical evaluations, physical examinations, and genetic testing.



Behrad Heidari, living with Duchenne muscular dystrophy. Picture by Duchenne Parent Project Netherlands

3 HOW DO YOU CARE FOR A PERSON WITH DUCHENNE MD?

- The current standard of care for DMD includes treatment with corticosteroids, giving physical therapy, taking care of the heart, and should be provided by a multidisciplinary team.
- The 2018 Care Considerations provide clinicians with the Standard of Care to be implemented when caring for people living with DMD.
- People with DMD need to be extra careful if they have a fall or bump into something because they are at a higher risk for Fat Embolism Syndrome (FES) in case of a fracture.

4 IS THERE A CURE OR TREATMENT FOR DUCHENNE MD?

- There are currently 7 (conditionally) approved treatments for DMD that slow down disease progression.
- There is no definitive 'cure' for DMD. Muscle tissue that is lost cannot be restored.
- Research efforts are focused on developing disease-modifying therapies, for example gene therapy, exon skipping, and gene editing techniques, to restore or add the missing or defective dystrophin protein.

