

Duchenne Muscular Dystrophy (DMD)



1. What is DMD?

Duchenne Muscular Dystrophy is a genetic disorder in which the body's muscles progressively degenerate and weaken. People with DMD may experience difficulty rising off the floor, maintaining balance, climbing staircases, and raising their arms. The disease most commonly affects males but in some rare cases it can affect females. Muscle weakness can present itself as early as age 3.

2. What Causes DMD?

In 1986, researchers found that when a particular gene on the X chromosome is mutated, there is likely to be a deficiency of the dystrophin protein which causes muscles to become weak and damaged. When a mother is a carrier of the gene, DMD could be inherited by her son. If the carrier mother has a daughter, the daughter will likely be a carrier of DMD as well.

3. How is DMD diagnosed?

Once muscle weakness is reported to a doctor, which should prompt diagnostic testing. The first test is a blood test called Creatine Kinase (CK) test. High Levels of CK suggests that the muscles are causing the weakness, not the nervous system. Next, DNA testing is available for the child and/or their mother. Typically, the life expectancy is shorter for people with DMD but research is expanding on disease-modifying therapies to increase life expectancy.

4. What are the main concerns for parents and families of children with DMD?

Early diagnosis and keeping up and being informed about medical care for a child with DMD could be challenging for a parent. Maintaining a positive attitude and maintaining a balanced family life is important to the parent and child. Empowering the child to be involved in the community and have a healthy psychosocial life will better the life of the child. For a mother this diagnosis can be devastating at first, recognizing and accepting the fact that it is not the mothers *fault* that their child has a disability is one of the first steps in moving forward to provide the best life possible for the child and the family as a whole.

For more information, visit:

- www.parentprojectmd.org
- <https://www.mda.org/disease/duchenne-muscular-dystrophy>
- <http://www.cdc.gov/ncbddd/musculardystrophy/facts.html>
- http://www.parentprojectmd.org/site/PageServer?pagename=nws_index
- http://www.uppmd.org/?option=com_frontpage&Itemid=1
- <http://www.healthynj.org/diseases/musculardystrophy.html>
- <http://www.ryansquest.org/>
- <https://www.facebook.com/parentprojectmd>