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adding value to lives

Focus on... Muscular Dystrophy

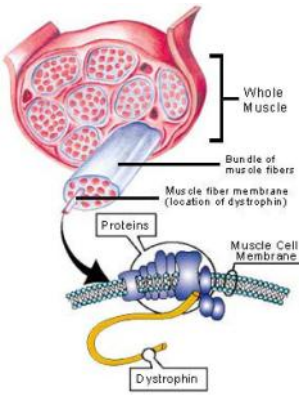
What is muscular dystrophy?

Muscular dystrophy (MD) is a broad term that describes a genetic (inherited) disorder of the muscles. Muscular dystrophy causes the muscles in the body to become very weak. The muscles break down and are replaced with fatty deposits over time.

Other health problems commonly associated with muscular dystrophy include the following:

- heart problems
- scoliosis (a condition that causes the back bones to curve)
- obesity.

The most common form of muscular dystrophy is called Duchenne's muscular dystrophy (DMD). Duchenne's muscular dystrophy usually affects only males. It occurs in one out of 3,500 live male births. Muscular dystrophy rarely affects girls, and when it does the condition is normally not as severe.



What causes muscular dystrophy?

Duchenne's muscular dystrophy is a genetic disease, which means it is inherited. Our genes determine our traits, such as eye color and blood type. Genes are contained in the cells of our bodies on stick-like structures called chromosomes. There are normally 46 chromosomes in each cell of our body, or 23 pairs. The first 22 pairs are shared in common between males and females, while the last pair determine gender and are called the sex chromosome pair: females have two X chromosomes, while males have one X and one Y chromosome.

Duchenne's muscular dystrophy is caused by an X-linked recessive gene. For this reason, a woman can carry a recessive gene on one of the X chromosomes unknowingly, and pass it on to a son, who will express the trait or disease. Therefore, half of the daughters have the gene and can pass it to the next generation. The other half do not have the gene and therefore cannot pass it on. Half of the sons do not have the gene and cannot pass it on. The other half of the sons have inherited the gene and will express the trait or disorder (in this case, DMD).

What are the symptoms of muscular dystrophy?

Muscular dystrophy is usually seen in children before the age of 5, with weakness typically affecting the shoulder and pelvic muscle as one of the initial symptoms. The following are the most common symptoms of muscular dystrophy. However, each child may experience symptoms differently. Symptoms may include:

- clumsy movement
- difficulty climbing stairs
- frequently trips and falls
- unable to jump or hop normally
- tip toe walking.

How is muscular dystrophy diagnosed?

The diagnosis of muscular dystrophy is made with a physical examination and diagnostic testing by the child's physician. During the examination, the child's physician obtains a complete prenatal and birth history of the child and asks if other family members are known to have muscular dystrophy. Diagnostic tests for muscular dystrophy may include:

- blood tests
- muscle biopsy - the primary test used to confirm diagnosis. A small sample of muscle tissue is taken and examined under a microscope.
- electromyogram (EMG) - test to check if the muscle weakness is a result of destruction of muscle tissue rather than nerve damage.
- electrocardiogram (ECG or EKG) - a test that records the electrical activity of the heart, shows abnormal rhythms (arrhythmias or dysrhythmias) and detects heart muscle damage.

Treatment for muscular dystrophy:

Specific treatment for muscular dystrophy will be determined by the child's physician based on:

- the child's age, overall health, and medical history
- the extent of the condition
- the type of condition
- the child's tolerance for specific medications, procedures, or therapies
- expectations for the course of the condition
- The parents' opinion or preference.

To date, there is no known treatment, medicine, or surgery that will cure muscular dystrophy, or stop the muscles from weakening. The goal of treatment is to prevent deformity and allow the child to function as independently as possible.

Since muscular dystrophy is a life-long condition that is not correctable, management includes focusing on preventing or minimizing deformities and maximizing the child's functional ability at home and in the community.

Management of muscular dystrophy is either non-surgical or surgical. Non-surgical interventions may include:

- physical therapy
- positioning aids - used to help the child sit, lie, or stand
- braces and splints - used to prevent deformity, promote support, or provide protection
- medications
- nutritional counseling
- psychological counseling.

Surgical interventions may be considered in some cases.

Long-term outlook

Muscular dystrophy is a progressive condition that needs life-long management to prevent deformity and complications. Walking and sitting often becomes more difficult as the child grows. Usually by the age of 12, the child needs a wheelchair because the leg muscles are too weak to work. Heart or lung problems often occur by the late teenage years or into the early 20s.