Muscular Dystrophy

DEFINITION

Muscular dystrophy is a group of genetic diseases in which muscle fibers are unusually susceptible to damage. These damaged muscles become progressively weaker. Most people who have muscular dystrophy will eventually need to use a wheelchair.

There are many different kinds of muscular dystrophy. Symptoms of the most common variety begin in childhood, primarily in boys. Other types of muscular dystrophy don't surface until adulthood.

People who have muscular dystrophy may have trouble breathing or swallowing. Their limbs may also draw inward and become fixed in that position — a problem called contracture. Some varieties of the disease can also affect the heart and other organs.

While there is no cure for muscular dystrophy, medications and therapy can slow the course of the disease.

(Source: Reprinted from the MayoClinic.com article "Muscular dystrophy: Definition" http://www.mayoclinic.com/health/muscular-dystrophy/DS00200)

SYMPTOMS

Progressive muscle weakness is the main feature of muscular dystrophy. Each separate form of muscular dystrophy varies a bit in terms of the age at which the signs and symptoms usually begin and the sequence in which different muscle groups are affected.

Duchenne muscular dystrophy

About half of all muscular dystrophy cases are the Duchenne variety, which most commonly occurs in boys. Signs and symptoms typically first surface when the child begins to walk and may include:

- Frequent falls
- Difficulty getting up from a lying or sitting position
- Trouble running and jumping
- Waddling gait
- Large calf muscles
- Learning disabilities

Becker muscular dystrophy

This variety has signs and symptoms similar to Duchenne muscular dystrophy, but they typically are milder and progress more slowly. Symptom onset is generally in the teens but may not occur until the mid-20s or even later.

Other types of muscular dystrophy

Certain other types of muscular dystrophy are defined by a specific feature or the location of the body where symptoms first begin. Examples include:

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- Myotonic. Also known as Steinert's disease, this form of muscular dystrophy also features an inability to relax muscles at will. It most often begins in early adulthood. Muscles of the face are usually the first to be affected.
- Limb-girdle. The hip and shoulder muscles are usually the first affected in this type of muscular dystrophy. In some cases, it becomes difficult to lift the front part of the foot, so frequent tripping may occur. Signs and symptoms may begin from early childhood to adulthood.
- Congenital. This category of muscular dystrophy is apparent at birth or becomes evident before age 2. Some forms progress slowly and cause only mild disability, while others progress rapidly and cause severe impairment.
- Fascioscapulohumeral (FSHD). One of the most striking signs of this variety of muscular dystrophy is that the shoulder blades might stick out like wings when the person raises his or her arms. Onset usually occurs in teens or young adults.
- Oculopharyngeal. The first sign of this type of muscular dystrophy is usually drooping of the eyelids. Weakness of the muscles of the eye, face and throat often results in swallowing difficulties. Signs and symptoms first appear in adulthood, usually in a person's 40s or 50s.

When to see a doctor

Seek medical advice if you notice signs of muscle weakness — such as increased clumsiness and falling — in yourself or your child.

(Source: Reprinted from the MayoClinic.com article " Muscular dystrophy: Symptoms" http://www.mayoclinic.com/health/muscular-dystrophy/DS00200/DSECTION=symptoms)

CAUSES

Hundreds of genes are involved in making proteins that protect muscle fibers from damage. Muscular dystrophy occurs when one of these genes is defective. Each form of muscular dystrophy is caused by a genetic mutation that's particular to that type of the disease. Many of these mutations are inherited, but some occur spontaneously in the mother's egg or the developing embryo.

(Source: Reprinted from the MayoClinic.com article " Muscular dystrophy: Causes" http://www.mayoclinic.com/health/muscular-dystrophy/DS00200/DSECTION=causes)

RISK FACTORS

Muscular dystrophy occurs in both sexes and in all ages and races, but the most common variety usually occurs in young boys. People who have a family history of muscular dystrophy are at higher risk of developing the disease or passing it on to their children.

(Source: Reprinted from the MayoClinic.com article " Muscular dystrophy: Risk Factors" http://www.mayoclinic.com/health/muscular-dystrophy/DS00200/DSECTION=risk-factors)

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COMPLICATIONS

Some types of muscular dystrophy shorten the person's lifespan, often by affecting the muscles associated with breathing. Even with improved mechanical breathing assistance, people who have Duchenne muscular dystrophy — the most common type of muscular dystrophy — usually die of respiratory failure before they reach age 40.

Many types of muscular dystrophy can also reduce the efficiency of the heart muscle. If the muscles involved with swallowing are affected, nutritional problems may develop.

As muscle weakness progresses, mobility becomes a problem. Many people who have muscular dystrophy will eventually need to use a wheelchair. However, the prolonged immobility of joints associated with wheelchair use can worsen contractures, in which the limbs draw inward and become fixed in that position.

Contractures may also play a part in the development of scoliosis, a sideways curvature of the spine that further reduces lung efficiency in people who have muscular dystrophy.

(Source: Reprinted from the MayoClinic.com article “Muscular dystrophy: Complications” http://www.mayoclinic.com/health/muscular-dystrophy/DS00200/DSECTION=complications)