

Muscular Dystrophy

Muscular dystrophy is the term for several inherited diseases that gradually damage muscle tissue. Each of these diseases is caused by a different gene abnormality and therefore cause different symptoms. Some are present at birth, whereas others do not appear until childhood or even adulthood. Unfortunately, there is no cure for muscular dystrophy. Treatment can help to manage some of the complications.

What is muscular dystrophy?

Several different diseases may be called “muscular dystrophy.” However, they are actually unrelated diseases, caused by different gene abnormalities. The one thing they have in common is that they all cause gradual destruction of muscle tissue.

Some muscular dystrophies are already apparent at birth. Others don't appear until adolescence or even young adulthood. These diseases vary in how quickly they develop, the severity of the disability, and the expected change of survival. There is no cure for any form of muscular dystrophy. Treatments can help to maximize your child's functioning and quality of life and may help to slow the course of the disease.

What does it look like?

There are many different muscular dystrophies. Each is caused by a specific abnormal gene, and each has specific symptoms. The three most common muscular dystrophies are Duchenne, Becker, and myotonic muscular dystrophy.

- *Duchenne muscular dystrophy.* Muscle weakness first appears in early childhood, usually by age 6. An abnormal walking pattern (gait) is often the first sign.
 - This muscular dystrophy occurs only in boys, but the abnormal gene is carried by females (“X-linked”).
 - Intelligence is reduced, but most patients are not severely mentally handicapped.
 - Walking becomes increasingly difficult. Even with treatment, most patients lose the ability to walk by age 12.
 - Muscle weakness spreads and worsens. Weakness of the muscles used in breathing and swallowing makes it increasingly difficult to keep the airways clear. Limitations on how well the limbs can straighten, called contractures, occur. Contractures limit movement of the limbs and can cause abnormal curving of the spine (scoliosis).
 - Weakness of the heart muscle develops (cardiomyopathy). This may further reduce your child's activity level.

- *Becker's muscular dystrophy.* Similar to Duchenne muscular dystrophy, but it develops later and is usually less severe.
 - Also occurs only in boys: first symptoms usually appear in the late teens or early adulthood.
 - Fewer learning problems than in Duchenne muscular dystrophy.
 - The pattern of increasing muscle weakness is similar to that in Duchenne muscular dystrophy but slower to develop. However, the same types of disabilities eventually appear. Many patients die before age 40.
- *Myotonic muscular dystrophy* (sometimes called Steinert's disease) affects various types of muscle. Other problems may include diseases of the heart, thyroid gland, and eyes (cataracts).
 - Muscle weakness usually develops around age 5. Affected infants may have a typical facial appearance. A severe newborn form can rapidly cause death.
 - The first sign is “myotonic” muscle contractions. This means that the muscles are very slow to relax after being used. The muscles of the fingers and face are affected first, other muscles later on. Speech may be affected because of weakening of the tongue and facial muscles.
 - About half of patients have reduced intelligence but are usually not severely mentally handicapped. The rest have normal intelligence.
 - Muscle weakness spreads very slowly; most patients can still walk even in late adulthood.

Many other types of muscular dystrophy are possible, each with its own pattern of onset and severity. Some types develop early and progress rapidly. Other forms develop later in life, develop slowly, and have little or no impact on life expectancy. Testing is an important first step to determine what type of muscular dystrophy your child has.

What causes muscular dystrophy?

Each form of muscular dystrophy is caused by a specific genetic (inherited) abnormality. The gene defects are already present at birth, even if muscle weakness and other symptoms don't develop until later.

What are some possible complications of muscular dystrophy?

All forms of muscular dystrophy cause progressive breakdown and destruction of muscle fibers. Some forms involve other parts of the body. The exact complications

caused by this muscle degeneration depend on which form your child has.

What increases your child's risk of muscular dystrophy?

- Duchenne muscular dystrophy occurs in about 1 in 3600 newborn boys. Myotonic muscular dystrophy affects about 1 in 30,000 infants.
- The genes for muscular dystrophy run in families. Some, like the gene abnormalities that cause Duchenne and Becker muscular dystrophies, are “X-linked.” This means that the gene is carried by females but the disease appears only in males. *Genetic counseling* can help you to understand this risk.

How is muscular dystrophy diagnosed?

- Your child will probably be referred to a *geneticist* (a specialist in inherited diseases) or a *neurologist* (a specialist in diseases of the nervous system). Besides performing tests to diagnose the disease, they will explain the risk of it occurring in children you or other family members may have in the future.
- *Genetic testing.* Some form of genetic testing is needed to identify the gene abnormality causing your child's disease. The same tests may be performed on the parents and other family members. This will help to determine where the abnormal gene came from and to identify family members who might pass it on to future children.
- *Muscle biopsy.* A sample of your child's muscle tissue may be obtained for examination under the microscope.

How is muscular dystrophy treated?

Unfortunately, there is no cure for muscular dystrophy. However, treatments are available to help keep your child

functioning as well as possible and to prevent and manage complications.

Treatments for muscular dystrophy depend on your child's specific medical problems. Physical therapy may help to keep muscles working as long as possible. Braces and other aids may be used to assist your child's mobility. Surgery is sometimes recommended to help preserve or improve your child's functioning. If the heart is involved, medications may be needed.

Regular medical follow-up is needed to preserve your child's heart and lung function as much as possible. Close attention to issues like diet and immunizations can help to preserve your child's health.

Mental health care and support are also important. Depression can be a problem, especially for adolescents with muscular dystrophy. Having a child with such a serious disease can be overwhelming for parents. Several national and local organizations are available to provide information and support for children with muscular dystrophy and their families.

Where can I get more information about muscular dystrophy?

Here are a few contacts to get you started in learning what your family needs to know about living with muscular dystrophy:

- The National Institute of Neurological Disorders and Stroke. Information and publications are available on the Internet at www.ninds.nih.gov.
- The Muscular Dystrophy Association. On the Internet at www.mdausa.org or call 1-800-FIGHT-MD (1-800-344-4863).