Muscular Dystrophy

Fact Sheet

What is Muscular Dystrophy?

Muscular dystrophy is a term that refers to a group of muscle disorders in which the face, arm, leg, spine, or heart muscles gradually shrink and weaken over time. There are a variety of different types of muscular dystrophy, including Duchenne muscular dystrophy, myotonic dystrophy, Becker's muscular dystrophy, Facioscapulohumeral muscular dystrophy, the limb-girdle muscular dystrophies, and the mitochondrial myopathies. The different types are distinguished by factors such as the:

- age at which symptoms usually start
- pattern of muscle weakness
- speed at which the disease progresses
- involvement of other tissues besides muscle
- pattern of inheritance

Muscular dystrophy is rare. For example, although Duchenne muscular dystrophy is one of the more common types, it occurs in only 1 out of every 50,000. The other types of muscular dystrophy are even more rare.

How is it manifested?

All muscular dystrophies are inherited. Each type of muscular dystrophy is associated with a distinct genetic mutation. The nature of the gene mutation and which chromosome it is located on determine the characteristics of the muscular dystrophy and the way the disease is passed from one generation to the next. The symptoms and age of onset depend on the type of muscular dystrophy. Symptoms of muscular dystrophy often include:

- problems with coordination and mobility with frequent falls
- muscle weakness
- joint stiffness

Duchenne muscular dystrophy symptoms appear early, usually between the ages of one and six. The condition progresses quickly, with many people needing a wheelchair by the age of 12. Symptoms include those listed above, plus problems with the bones in the back and chest, as well as fatigue. Those with the condition may eventually have trouble breathing and develop pneumonia. They may also sometimes grow to be mentally challenged. Becker's muscular dystrophy, which usually starts around age 12, is much less severe than Duchenne muscular dystrophy. Symptoms include those listed above, plus heart disease, problems with the curvature of the spine, fatigue, problems with thinking, and breathing difficulties. Symptoms of facioscapulohumeral muscular dystrophy (also called Landouzy-Dejerine muscular dystrophy) include weak facial and shoulder muscles and difficulty with lifting.
arms, whistling, and closing eyes. It affects mainly the upper body and can also cause hearing loss, speech problems, and changes in heart rhythm. Symptoms often start around ages 10 to 26 years old, but may also start at a later stage. **Limb-girdle muscular dystrophies** cause weakness in the hips or shoulders. Symptoms usually start to appear in childhood or early adulthood. Muscle weakness starts in the pelvic area, and then moves to the shoulders and other body parts. **Mitochondrial myopathies** are caused by defects in the genes of the mitochondria, which are specialized units found inside cells that create the energy needed for cells to work. Symptoms include those listed above, plus heart problems, seizures, and problems with vision and hearing. **Myotonic muscular dystrophy** usually does not start to produce symptoms until after the age of 20. Some people don't have symptoms until as late as age 50. This type of muscular dystrophy can also affect the heart, eyes (cataracts), lungs, digestive tract, and brain function and can be associated with diabetes. There is, however, a distinct difference between the type that affects newborn infants — congenital MMD — and the type that begins in adolescence or adulthood — adult-onset MMD. Infants with congenital MMD have severe muscle weakness, including weakening of the muscles that control breathing and swallowing. These problems can be life threatening and need intensive care.

**How is it diagnosed or detected?**

In diagnosing any form of muscular dystrophy, a doctor usually begins by taking a patient and family history and performing a physical examination. Much can be learned from these, including the pattern of weakness. The history and physical go a long way toward making the diagnosis, even before any complicated diagnostic tests are done. The doctor also wants to determine whether the patient's weakness results from a problem in the muscles themselves or in the nerves that control them. Problems with muscle-controlling nerves, or motor nerves, originating in the spinal cord and reaching out to all the muscles, can cause weakness that looks like a muscle problem but really isn't. Usually, the origin of the weakness can be pinpointed by a physical exam. Occasionally, special tests called nerve conduction studies and electromyography (EMG) are done. In these tests, electricity and very fine pins are used to stimulate and assess the muscles or nerves individually to see where the problem lies. Electromyography is uncomfortable but not usually very painful.

**Who is affected?**

MD occurs worldwide, affecting all races. Its incidence varies, as some forms are more common than others. Its most common forms in children, Duchenne and Becker muscular dystrophy, alone affect approximately 1 in every 3,500 to 5,000 boys, or between 400 and 600 live male births each year (United Stated source).

**Additional Resources:**

**Muscular Dystrophy Canada** - [www.muscle.ca](http://www.muscle.ca) National Office: Muscular Dystrophy Canada 2345 Yonge St, Suite 900 Toronto, Ontario M4P 2E5 Telephone: 1-866-MUSCLE-8 Muscular Dystrophy Canada’s dedicated volunteers and staff across the country raise funds and work hard to support the independence and full participation of Canadians with neuromuscular disorders, fund research to find a cure and improve the quality of life of people with...
neuromuscular disorders, assist our clients to participate in the decisions that affect them, and collaborate with others for social change.

Books and Literature:

Author: Irwin M. Siegel This book covers everything from available medical treatments to helping the child grow up with a positive self-image to what the future holds for the treatment of muscular dystrophy and more. Muscular Dystrophy: The Facts (ISBN-13: 978-0192632173) Author: Alan Emery Written specifically for people with muscular dystrophy and their families, this new edition of Muscular dystrophy: the facts answers many of the questions asked about how and why it occurs, and how it will affect the life of a recently diagnosed child. Throughout, the different types of muscular dystrophy are described with a minimum of technical jargon. The content contained in this document is for general information purposes. It is not intended to diagnose or treat a child.