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Muscular Dystrophy

Aside from seeing the telethon hosted by Jerry Lewis on Labor Day weekend, many people don't know much about muscular dystrophy. Yet a quarter of a million kids and adults are living with the disease, so chances are you may know someone who has it.

What Is Muscular Dystrophy?

Muscular dystrophy (MD) is a genetic disorder that weakens the muscles that help the body move. People with MD have incorrect or missing information in their genes, which prevents them from making the proteins they need for healthy muscles. Because MD is genetic, people are born with the problem — it's not contagious and you can't catch it from someone who has it.

MD weakens muscles over time, so children, teens, and adults who have the disease can gradually lose the ability to do the things most people take for granted, like walking or sitting up. Someone with MD might start having muscle problems as a baby or their symptoms might start later. Some people even develop MD as adults.

Several major forms of muscular dystrophy can affect teens, each of which weakens different muscle groups in various ways:

- **Duchenne** (pronounced: due-**shen**) **muscular dystrophy (DMD)**, the most common type of the disease, is caused by a problem with the gene that makes a protein called **dystrophin**. This protein helps muscle cells keep their shape and strength. Without it, muscles break down and a person gradually becomes weaker. DMD affects boys. Symptoms usually start between ages 2 and 6. By age 10 or 12, kids with DMD often need to use a wheelchair. The heart may also be affected, and people with DMD need to be followed closely by a lung and heart specialist. They can also develop scoliosis (curvature of the spine) and tightness in their joints. Over time, even the muscles that control breathing get weaker, and a person might need a ventilator to breathe.
- **Becker muscular dystrophy (BMD)**, like DMD, affects boys. The disease is very similar to DMD, but its symptoms may start later and can be less severe. With BMD, symptoms like muscle breakdown and weakness sometimes don't begin until age 10 or even in adulthood. People with BMD can also have breathing, heart, bone, muscle, and joint problems. Many people with BMD can live long, active lives without using a wheelchair.
- **Emery-Dreifuss** (pronounced: **em**-uh-ree **dry**-fuss) **muscular dystrophy (EDMD)** typically starts causing symptoms in late childhood to early teens and sometimes as late as age 25. EDMD is

another form of muscular dystrophy that affects mostly boys. It involves muscles in the shoulders, upper arms, and shins, and it often causes joint problems (joints can become tighter in people with EDMD). The heart muscle may also be affected.

- **Limb-girdle muscular dystrophy (LGMD)** affects boys and girls equally, weakening muscles in the shoulders and upper arms and around the hips and thighs. LGMD can begin as early as childhood or as late as mid-adulthood, and it often progresses slowly. Over time, a wheelchair might be necessary to get around. There are many different types of LGMD, each with its own specific features.
- **Facioscapulohumeral** (pronounced: **fa-she-o-skap-you-lo-hyoo-meh-rul**) **muscular dystrophy (FSHD)** can affect both guys and girls, and it usually begins during the teens or early adulthood. FSHD affects muscles in the face and shoulders and sometimes causes weakness in the lower legs. People with this type of MD might have trouble raising their arms, whistling, or tightly closing their eyes. How much a person with this form of muscular dystrophy is affected by the condition varies from person to person. It can be quite mild in some people.
- **Myotonic** (pronounced: **my-uh-tah-nick**) **dystrophy (MMD)** is a form of muscular dystrophy in which the muscles have difficulty relaxing. In teens, it can cause a number of problems, including muscle weakness and wasting (where the muscles shrink over time), cataracts, and heart problems.
- **Congenital muscular dystrophy (CMD)** is the term for all types of MD that show signs in babies and young children, although the MD isn't always diagnosed right away. Like other forms of MD, CMD involves muscle weakness and poor muscle tone. Occurring in both girls and boys, it can have different symptoms. It varies in how severely it affects people and how quickly or slowly it worsens. In rare cases, CMD can cause learning or intellectual disabilities.

The life expectancy (in other words, how long a person may live) for many of these forms of muscular dystrophy depends on the degree to which a person's muscles are weakened as well as how much the heart and lungs are affected.

How Do Doctors Diagnose MD?

In addition to doing a physical examination, the doctor will ask you about any concerns and symptoms you have, your past health, your family's health, any medications you're taking, any allergies you may have, and other issues. This is called the **medical history**.

Tests can help the doctor determine which type of MD a person has and rule out other diseases that affect the muscles or nerves. Some tests measure how nerves and muscles are working. Others check the blood for levels of certain **enzymes**, the proteins that cause chemical changes like converting food to energy. Abnormally high blood levels of certain enzymes from muscle cells are present in many people with MD. Blood tests are available to diagnose some of these disorders. They are genetic tests that look at the DNA. These tests can be useful in diagnosing Duchenne and Becker muscular dystrophy, certain limb girdle muscular dystrophies, Fascioscapular humeral muscular dystrophy, myotonic dystrophy, Emery Dreifus

muscular dystrophy, ocular pharyngeal muscular dystrophies and certain congenital muscular dystrophies.

Sometimes a **muscle biopsy** is needed. The doctor removes a small piece of muscle tissue and examines it under a microscope. If a person has MD, the muscle tissue will have some unusually large fibers, and some of the other fibers will show signs of breaking down. Finally, genetic testing can show if a person has Duchenne MD or certain other forms of muscular dystrophy.

How Is MD Treated?

There is no cure for MD, but doctors and scientists are working hard to find one. Some scientists are trying to fix the defective genes that lead to MD so they will make the right proteins. Others are trying to make chemicals that will act like these proteins in the body. They hope that this will help the muscles to work properly in people with MD. Doctors are also dedicated to finding the best ways to treat the symptoms of MD so that kids, teens, and adults with the disease can live as comfortably and happily as possible.

Teens with MD can do some things to help their muscles. Certain exercises and physical therapy can help them avoid **contractures**, a stiffening of the muscles near the joints that can make it harder to move and can lock the joints in painful positions. Often, teens are fitted with special braces to ensure flexible joints and **tendons** (the strong, rubber band-like tissues that attach muscles to bones). Surgery is sometimes used to reduce pain and increase movement from contractures.

Because we rely on certain muscles to breathe, some teens with MD need respiratory aids, such as a ventilator, to help them breathe. Teens with MD also might need to be treated for problems like scoliosis, which can be caused by weakened muscles or muscles that are contracting or pulling too tightly.

For some types of MD, medication can help. Guys with Duchenne MD may be helped by a medicine called prednisone, and teens with myotonic MD might use mexilitine to relax muscles.

It's also important that people with heart problems caused by muscular dystrophy be monitored by a heart specialist.

What's It Like for Teens With MD?

Teens have different experiences depending on the type of MD. One person might have weakened shoulder muscles and not be able to raise a hand in class. Someone might be unable to smile because of weak facial muscles. Another person might have weak muscles in the pelvis or legs, making it hard to walk from class to class. In some cases, you might not even be able to tell that a teen has MD.

For teens with MD, it can be hard to come to terms with the disease, especially because it gradually gets worse. For example, when someone who walks to class must start using a wheelchair in school, it can be a difficult adjustment. Support from doctors, family, and friends can make it easier to deal with MD. Changes like wider doorways at home and school can make it easier for teens with MD to do many of the things they enjoy.

If you know someone who has MD, there's a lot you can do to offer help and support. For example, some people with MD may need help getting books out during class or rides to and from events.

Often, people with illnesses that gradually get worse over time can start to feel cut off from friends, especially as their friends may be going out and doing more things while they feel like they're becoming more housebound. Try planning activities that include a friend, brother, or sister with MD, such as playing video games or watching movies at his or her home. Your friend or sibling will always be the same person — just more limited in movement.

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