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en español

Distrofia muscular

What Is Muscular Dystrophy?

Muscular dystrophy (MD) is a genetic disorder that gradually weakens the body's muscles. It's caused by incorrect or missing genetic information that prevents the body from making the proteins needed to build and maintain healthy muscles.

A child who is diagnosed with MD gradually loses the ability to do things like walk, sit upright, breathe easily, and move the arms and hands. This increasing weakness can lead to other health problems.

There are several major forms of muscular dystrophy, which can affect the muscles to varying degrees. In some cases, MD starts causing muscle problems in infancy; in others, symptoms don't appear until adulthood.

There is no cure for MD, but researchers are quickly learning more about how to prevent and treat it. Doctors are also working on improving muscle and joint function and slowing muscle deterioration so that those with MD can live as actively and independently as possible.

What Are the First Symptoms of Muscular Dystrophy?

Many kids with muscular dystrophy follow a normal pattern of development during their first few years of life. But in time they develop problems with movement. A child who has MD may start to stumble, waddle, have difficulty going up stairs, and toe walk (walk on the toes without the heels hitting the floor). A child may start to struggle to get up from a sitting position or have a hard time pushing things, like a wagon or a tricycle.

Kids with MD often develop enlarged calf muscles (called **calf pseudohypertrophy**) as muscle tissue is destroyed and replaced by fat.

How Is Muscular Dystrophy Diagnosed?

When first suspecting that a child has muscular dystrophy, a doctor will do a physical exam, take a family history, and ask about any problems — particularly those affecting the muscles — that the child might be having.

In addition, the doctor may do tests to see what type of MD is involved and to rule out other diseases that could cause the problem. These might include a blood test to measure levels of **serum creatine kinase**, an enzyme that's released into the bloodstream when muscle fibers are breaking down. High levels indicate that something is causing muscle damage.

The doctor also may do a blood test to check the DNA for gene defects or do a muscle biopsy. In a muscle biopsy, the doctor surgically removes a small sample of muscle and looks at it under the microscope. The muscle cells of a person with MD have changes in

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different degrees of muscle weakness.

- **Duchenne muscular dystrophy,** the most common form of muscular dystrophy. [illegible] body's muscles weaker.
- [illegible] s a Duchenne MD.
- **Myotonic dystrophy**, also known as Steinert's disease, is the most common *adult* form of MD, although half of all cases are diagnosed in people under 20 years old. The main symptoms include muscle weakness, myotonia (in which the muscles have trouble relaxing once they contract), and muscle wasting (when the muscles shrink over time).
- **Limb-girdle muscular dystrophy** affects boys and girls equally. Symptoms usually start when kids are between 8 and 15 years old. This form progresses slowly, affecting the pelvic, shoulder, and back muscles. The severity of muscle weakness varies — some kids have only mild weakness while others develop severe disabilities and as adults need to use a wheelchair.
- **Facioscapulohumeral muscular dystrophy** can affect both boys and girls, and the symptoms usually first appear during the teen years. It tends to progress slowly. Muscle weakness first develops in the face, making it difficult for a child to close the eyes, whistle, or puff out the cheeks. The shoulder and back muscles gradually become weak, and kids have trouble lifting objects or raising their hands overhead. Over time, the legs and pelvic muscles also may lose strength.

Caring for a Child With MD

There's no cure for MD yet, but doctors are working to improve muscle and joint function, and slow muscle deterioration.

If your child is diagnosed with MD, a team of medical specialists will work with you and your family, including: a neurologist, orthopedist, pulmonologist, physical and occupational therapist, nurse practitioner, cardiologist, registered dietician, and a social worker.

Muscular dystrophy is often degenerative, meaning that it slowly affects more organs and can't be reversed. So, kids may pass through different stages as it progresses and need different kinds of treatment. During the early stages, physical therapy, joint bracing, and medications are often used.

During the later stages, doctors may use assistive devices, such as:

- physical therapy and bracing to improve flexibility
- power wheelchairs and scooters to improve mobility
- a ventilator to support breathing
- robotics to help with routine daily tasks

Physical Therapy and Bracing

Physical therapy can help a child maintain muscle tone and reduce the severity of joint contractures with exercises that keep the muscles strong and the joints flexible.

A physical therapist also uses bracing to help prevent joint contractures, a stiffening of the muscles near the joints that can make it harder to move and can lock the joints in painful positions. By providing extra support in just the right places, bracing can extend the time that a child with MD can walk independently.

Spinal Fusion

lessen the curvature so a child can sit upright and comfortably in a chair. This surgery ensures that the spine curvature doesn't have an effect on breathing. Usually, spinal fusion surgery only requires a short hospital stay.

Res

Many kids with muscular dystrophy also have weakened heart and respiratory muscles. As a result, they can't cough out phlegm and sometimes develop respiratory infections that can quickly become serious. Good general health care and regular vaccinations are especially important for children with muscular dystrophy to help prevent these infections.

Assistive Devices

A variety of new technologies can provide independence and mobility for kids with muscular dystrophy.

Some kids with Duchenne MD might use a manual wheelchair once it becomes difficult to walk. Others go directly to a motorized wheelchair, which can be equipped to meet their needs as muscle deterioration advances.

Robotic technologies also are under development to help kids move their arms and perform activities of daily living.

If your child would benefit from an assistive technological device, contact your local chapter of the Muscular Dystrophy Association to ask about financial help that might be available. In some cases, health insurers cover the cost of these devices.

The Search for a Cure

Researchers are quickly learning more about what causes the genetic disorder that leads to muscular dystrophy, and about possible treatments for the disease.

To learn more about the most current research on MD, contact the local chapter of the Muscular Dystrophy Association or talk to your doctor, who also can tell you about clinical trials on MD.

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