

# Duchenne Muscular Dystrophy

## What Is Duchenne Muscular Dystrophy?

Duchenne (due-shen) muscular dystrophy (DMD) is a genetic neuromuscular disorder that causes progressive muscle loss and weakness. These symptoms lead to serious medical problems, especially with the heart and lungs. The condition mostly affects males.

With advances in cardiac and respiratory care, more people who have DMD are living into their 30s, with some living into their 40s and 50s.

DMD affects roughly one in 3,600-5,000 male infants, in the U.S. and occurs in all races.

## What Causes Duchenne Muscular Dystrophy?

DMD disease occurs when a gene on the X chromosome changes (mutates) and fails to produce enough of the muscle-protecting protein dystrophin.

Because of the way DMD gets passed down from parent to child, it mostly affects boys. Every boy inherits an X chromosome from his mother and a Y chromosome from his father. The gene that can carry a mutation in dystrophin production is on the X chromosome.

DMD usually doesn't affect girls, because girls inherit two X chromosomes (one from each parent). When a girl inherits a mutated dystrophin gene from one parent, she also receives a healthy one from her other parent, protecting her from the disease. Because boys don't receive a second dystrophin-producing gene to balance out the mutated one, they are affected by DMD.

Female carriers typically don't have any symptoms of DMD, however up to 20 percent may show symptoms of cardiomyopathy or other muscle weakness. Sons of women who carry a mutated gene have a 50 percent chance of having DMD. Daughters of female carriers have a 50 percent chance of being DMD carriers.

DMD usually runs in families, but it's possible for a family with no history of the condition to have a son who has the disease or a daughter who's a carrier. Sometimes new dystrophin gene mutations occur in a mother's egg cells. Once a new mutation passes to a son or daughter, it can be passed to the next generation.

## Duchenne Muscular Dystrophy Symptoms and Effects

The symptoms and effects of DMD change as time goes by, so the progression of the condition is divided into stages.

The symptoms associated with each stage of DMD overlap, and differ for each child. DMD usually gets diagnosed between ages 2 and 7. Often, delays in early milestones—such as walking—show the first evidence of the condition.

### Infants to Preschoolers (0 to 5 Years)

Children in this age range who have DMD might:

- Move more slowly.
- Have trouble speaking.
- Have learning or behavior problems.
- Fall frequently.
- Get tired easily.
- Have fibrosis of the muscles. Fibrosis occurs when scar tissue replaces damaged muscles, causing joint tightness.

### School Age (6 to 10 Years)

Children in this age range who have DMD might:

- Start experiencing weakened arms, hands and thigh muscles (called quadriceps).
- Have difficulty walking, or shift their weight when walking.
- Need a stroller, scooter or manual wheelchair for longer distances.
- Get tired easily.

### Preteens and Younger Teens (11 to 15 Years)

Children in this age range who have DMD might:

- Not be able to walk.
- Need to use a power wheelchair.
- Be at risk for developing scoliosis.
- Experience additional weakness in the arms and hands.

### Older Teens and Adults (16 and Older)

Teens and adults in this age range who have DMD:

- Will likely need a power wheelchair.
- Might have some remaining function in their hands.
- Often have difficulty maintaining good posture.
- Might have difficulty breathing and need to use a respirator.

## **Duchenne Muscular Dystrophy Treatment and Diagnosis**

The following tests are used to diagnose DMD.

- **Physical exam:** A physical exam tests gross motor abilities.
- **Creatine kinase (CK) level:** This blood test measures levels of an enzyme called CK, which is produced by damaged muscles. High CK levels can mean that muscles are degenerating due to conditions such as muscular dystrophy.
- **Genetic testing:** This blood test reveals mutations in the gene responsible for DMD. Female relatives of boys who have DMD can get a genetic test to see if they're carriers.
- **Muscle biopsy test:** If needed, doctors remove a small sample (called a biopsy) of muscle tissue. They examine the sample under a microscope, looking for signs of muscle degeneration.

Although there's no cure for Duchenne muscular dystrophy, the right care can reduce complications and improve quality of life for your child. As soon as specialists at Gillette Children's make a diagnosis, our team works with your family to create custom care plans. Regular checkups are important to keep symptoms under control and avoid complications.

As your child reaches adulthood, we continue to provide age-appropriate care through **Gillette Lifetime** for teens (16 and older) and adults.

## Integrated Care

If your child has Duchenne muscular dystrophy, our team of internationally recognized specialists will work closely with you and your family to create a treatment plan that fits your unique needs. Through the **Muscular Dystrophy Association (MDA)** Care Center at Gillette you'll have access to additional services and support in your community.

Your child will receive care from one of the Twin Cities' largest groups of **pediatric neurologists**, **pediatric rehabilitation medicine specialists** and **pediatric orthopedic surgeons**. They might also work with experts in areas related to muscle disorders, including:

- **Orthotics, Prosthetics and Seating.**
- **Cardiology.**
- **Medical genetics and genetic counseling.**
- **Neurology.**
- **Neuropalliative care.**
- Nursing.
- **Orthopedics.**
- **Pulmonology and respiratory care.**
- **Rehabilitation medicine.**

- [Sleep medicine](#).

## Testing and Diagnostic Services

- [Electromyography](#) test.
- Electrocardiogram (EKG) test.
- Functional ability tests.
- Molecular genetic tests.
- Muscle functional and strength tests.
- Pulmonary and respiratory tests.
- [Seating evaluations](#).
- [Sleep studies](#) (polysomnography).
- [Swallowing studies](#) (fluoroscopy and videofluoroscopy).
- [X-rays](#).

## Rehabilitation Therapies

- [Nutrition and feeding](#).
- [Occupational therapy](#).
- [Physical therapy](#).
- [Speech and language therapy](#).

## Family Support

- [Child life specialists](#).
- [Neuropalliative care](#).
- [Neuropsychologists](#).
- [Psychologists](#).
- [Social workers](#).
- [Therapeutic recreation specialists](#).

## Explore Duchenne Muscular Dystrophy Resources

- Learn more about neuromuscular diseases and see current research at the [Muscular Dystrophy Association](#).

- Understand more about DMD at [Parent Project Muscular Dystrophy](#).
- Review recent [DMD research](#) at Gillette.
- See current research from the [Paul and Sheila Wellstone Muscular Dystrophy Center](#) and the [Wellstone Muscular Dystrophy Research Network](#).

## About MDA Care Centers

[Muscular Dystrophy Association \(MDA\)](#) cares for kids and adults from day one at our network of more than 150 MDA Care Centers across the United States and Puerto Rico. These state-of-the-art clinics, located at top hospitals and health care institutions, bring health care specialists from a variety of disciplines together so families receive the care they need at one time, in one place. MDA Care Centers receive nearly 50,000 visits each year while also serving at the forefront of research by hosting clinical trials for the latest promising therapies.

[Appointment: 651-290-8707](#) [Refer a Patient: 651-325-2200](#) [Pediatric Expert Consult](#) [More Ways to Contact Us](#)

This information is for educational purposes only. It is not intended to replace the advice of your health care providers. If you have any questions, talk with your doctor or others on your health care team.

If you are a Gillette patient with urgent questions or concerns, please contact Telehealth Nursing at [651-229-3890](#).