



Muscular Dystrophy (MD)

Muscular dystrophy literally means the wasting away or atrophy of muscles. The muscular dystrophies (MD) refer to the group of genetic diseases characterized by progressive weakness and degeneration of the skeletal muscles that control movement. There are many forms of muscular dystrophy, some noticeable at birth known as congenital muscular dystrophy while other forms develop in adolescence (Becker MD). Regardless of the exact timing of onset, some muscular dystrophies lead to mobility impairment or even paralysis.

Q: What causes muscular dystrophy?

Muscular dystrophy is caused by a genetic mutation of the genes responsible for muscle protein production, leading to missing or faulty proteins that cause progressive muscle weakness and breakdown, with most cases inherited from parents but some arising from new, spontaneous mutations.

Q: Are there different types of muscular dystrophy?

Yes, the three most common types of MD are: Duchenne, facioscapulohumeral, and myotonic. These three types differ in terms of pattern of inheritance, age of onset, rate of progression, and distribution of weakness.

The three most common types of Muscular Dystrophy are:

- Duchenne MD primarily affects boys and is the result of mutations in the gene that regulates dystrophin – a protein involved in maintaining the integrity of muscle fiber. Onset is between 3-5 years and progresses rapidly. Most boys become unable to walk at 12, and by 20 must use a respirator to breathe.
- Facioscapulohumeral MD appears in adolescence and causes progressive weakness in facial muscles and certain muscles in the arms and legs. It progresses slowly and can vary in symptoms from mild to disabling.
- Myotonic MD is a rare, multi-systemic, progressive, inherited disease that is estimated to affect as many as 1 in 2,100 people. Myotonic dystrophy is the most common form of adult muscular dystrophy and is considered the most variable of all known conditions. The symptoms become more severe with each generation – known as genetic anticipation – yet there is currently no cure and there are no FDA approved treatments.

Q: How is muscular dystrophy diagnosed?

Muscular dystrophy is generally diagnosed during a physical exam while discussing family history and any issues the patient may be having. Testing can be done in a variety of ways, including blood tests, muscle biopsies, and genetic testing. For more on diagnosis, please see:

<https://www.nichd.nih.gov/health/topics/musculardys/conditioninfo/diagnosed>.

Q: What are the symptoms/side effects of muscular dystrophy?

The symptoms can include muscle and movement-related symptoms such as muscle atrophy, difficulty walking, running or climbing stairs, stiff or loose joints, spasticity and/or tightening of the muscles. Other symptoms can include tiredness, scoliosis, heart issues, breathing issues, and intellectual disabilities or cognitive impairment.

Q: How is muscular dystrophy treated?

There is no specific treatment for any of the various forms of MD. Physical therapy is often used to help prevent painful muscle contractures. Additionally, certain prescribed

medications may be used for pain management as well as for arresting muscular deterioration within some forms of MD. Orthopedic appliances are used for support while corrective orthopedic surgery may be required to improve the quality of life for others. In some cases, respiratory therapy may be needed. Finally, cardiac abnormalities may require a pacemaker.

Q: What else should I know about myotonic dystrophy?

Myotonic dystrophy is caused by a mutation in the DMPK gene, resulting in myotonic dystrophy type one, and in the CNBP gene, resulting in myotonic dystrophy type two. These mutations prevent the genes from carrying out their functions properly, impacting multiple body systems.

The genetic mutation is an autosomal dominant mutation, where one copy of the altered gene is sufficient to cause the disorder. As a result, affected individuals have a 50% chance of passing on the mutated gene to their children. A child is equally likely to have inherited the mutated gene from either parent. If both parents do not have the disease, their children cannot inherit it.

Through this inherited genetic anomaly, individuals with myotonic dystrophy experience varied and complex symptoms, from skeletal muscle problems, to heart, breathing, digestive, hormonal, speech and swallowing, diabetic, immune, excessive daytime sleepiness, early cataracts and vision, and cognitive difficulties.

Myotonic dystrophy is a highly variable and complicated disorder. The systems affected, the severity of symptoms, and the age of onset of those symptoms vary greatly between individuals, even in the same family. In general, the younger an individual is when symptoms first appear, the more severe symptoms are likely to be.

A complete diagnostic evaluation, which includes family history, physical examination, and medical tests, is typically required for a presumptive diagnosis of myotonic dystrophy. The presence of the disorder can then be confirmed by genetic testing. Prenatal testing, where the DNA of the fetus is checked for the presence of the myotonic dystrophy mutation, is also available. Despite the availability of simple genetic tests, misdiagnoses persist for decades.

Delays in diagnosing myotonic dystrophy are common, sometimes over a decade long. This is usually because of the lack of familiarity with the disease on the part of clinicians and that more common diseases with symptoms that mimic myotonic dystrophy must typically first be ruled out before this disorder is considered.

Myotonic MD varies in the age of onset and is characterized by myotonia (prolonged muscle spasm) in the fingers and facial muscles: a floppy-footed, high-stepping gait; cataracts; cardiac abnormalities; and endocrine disturbances. Individuals with myotonic MD have long faces and drooping eyelids; men have frontal baldness.

Q: Are there any clinical trials for muscular dystrophy?

There are clinical trials for MD looking into symptom management, targeted therapies, and gene therapies. Generally, <https://clinicaltrials.gov/> is a good source of information for new and completed trials.

Sources: Cleveland Clinic <https://my.clevelandclinic.org/health/diseases/14128-muscular-dystrophy>, National Institutes of Health <https://www.nichd.nih.gov/health/topics/muscularmys/conditioninfo/diagnosed>, Myotonic Dystrophy Foundation

Need to talk to someone?

Our Information Specialists are available to answer your questions.
Call toll-free 1-800-539-7309 Mon-Fri, 7 am - 8 pm EST
or schedule a call or ask a question online at
<https://www.ChristopherReeve.org/Ask>.

Resources for Muscular Dystrophy:

Muscular Dystrophy Association (MDA)

<https://www.mda.org/>

National Headquarters

1016 W. Jackson Blvd., #1073

Chicago, IL 60607

Phone: 800-572-1717, 888-ASK-MDA1 (833-275-6321)

Email: ResourceCenter@mda.org

MDA offers services such as educational webinars, on-demand learning, community activities, support groups, peer connection opportunities, 30-minute video calls to discuss resources and support needed, MDA summer camp for children, partnerships with specialty clinics and clinicians, a National Resource Center, fundraising activities/events, and information about the latest research. For more information on their support groups, please visit: <https://www.mda.org/care/community-groups>.

Centers for Disease Control: Muscular Dystrophy

<https://www.cdc.gov/muscular-dystrophy/>

The CDC sponsors MD STARnet, the Muscular Dystrophy Surveillance Tracking and Research Network, a program set up in several states to identify all individuals who have Duchenne/Becker Muscular Dystrophy. <https://www.cdc.gov/muscular-dystrophy/research/index.html>

Coalition Duchenne

www.coalitionduchenne.org

3857 Birch Street. Suite 263

Newport Beach, CA 92660

Phone: 714-801-4616

Email: Catherine@coalitionduchenne.org

Coalition Duchenne is a non-profit organization that raises global awareness and funding for Duchenne muscular dystrophy research through donations and various annual fundraising events.

CureDuchenne

<http://www.cureduchenne.org/>

100 Bayview Circle, Suite 5600

Newport Beach, CA 92660

Phone: 949-872-2552

Email: info@cureducheene.org

CureDuchenne's goal is to identify research with the most likelihood of making it to clinical trials and then provide the financial bridge that will take it from the lab and into human trials

Duchenne and You

www.DuchenneAndYou.com

Offers information and resources on Duchenne

Duchenne Registry (formerly Duchenne Connect)

<https://www.duchenneregistry.org/>

This site provides news and a registry for Duchenne MD.

Duchenne Registry

<https://www.duchenneregistry.org/>

This site provides a registry for those living with Duchenne muscular dystrophy.

Find-a-Cure for Children with Duchenne, Inc.

<http://www.findacure.com/>

E-mail: findacure@comcast.net

Find-a-Cure funds research to find a cure for Duchenne muscular dystrophy.

Hereditary Neuropathy Foundation (HNF)

<http://www.hnf-cure.org/>

401 Park Ave., #10

New York, NY 10016

Phone: 919-824-7260

E-mail: info@hnf-cure.org

HNF is a non-profit organization which raises awareness, funds scientific research, and educates the medical community as well as the general public about Charcot-Marie-Tooth disease (CMT).

Jett Foundation Inc.

<https://www.jettfoundation.org/>

390 Circuit St.
Norwell, MA 02061
Phone: 781-585-5566

Email: info@jettfoundation.org

This foundation is dedicated to funding Duchenne muscular dystrophy research. Its primary focus is direct service programming for families impacted by Duchenne and other neuromuscular disorders. They host a weeklong camp, family workshops, community seminars, and more. They also have the Jett Giving Fund, which offers families financial assistance through three branches of support: the Accessible Vehicle Fund, Equipment Assistance Fund, and Emergency Fund.

KidsHealth: Muscular Dystrophy

<https://kidshealth.org/en/parents/muscular-dystrophy.html?ref=search>

This page has information on muscular dystrophy written for children.

Medline Plus: Muscular Dystrophy

<http://www.nlm.nih.gov/medlineplus/musculardystrophy.html>

This page has information on muscular dystrophy including diagnosis, treatment, and coping.

Muscular Dystrophy Family Fund

<http://www.mdff.org/>

PO Box 776
Carmel, IN 46082
Phone: 317-615-9140

MDF exists to provide resources, services, and adaptive equipment to enable patients with muscular dystrophy and their family members to live independent and productive lives.

Myotonic Dystrophy Foundation (MDF)

<https://www.myotonic.org>

663 Thirteenth St., Suite 100
Oakland, CA 94612
Phone: 415-800-7777

Email: info@myotonic.org

The Myotonic Dystrophy Foundation (MDF) provides comprehensive programs and resources to support individuals and families affected by myotonic dystrophy (DM). MDF connects community members through virtual and in-person support groups, offers one-on-one assistance via the MDF Warmline, and maintains a Find-a-Doctor directory to help locate clinicians referred by people living with DM or their caregivers. Education and connection are supported through the annual MDF Conference, Regional Conferences, and the MDF Digital Academy, an on-demand library of expert presentations and lived-experience discussions.

To help families navigate care, MDF publishes evidence-based clinical care guidelines for both DM1 and DM2, an Anesthesia Quick Reference Guide for surgical teams, and

practical resources such as the Mental Health Handbook and system-specific care guides. These tools empower individuals to advocate for their own care and share critical information with healthcare providers. Together, MDF's programs and resources build knowledge, connection, and confidence for everyone affected by myotonic Dystrophy.

National Institutes of Neurological Disorders and Stroke (NINDS): Muscular Dystrophy Information Page

<https://www.ninds.nih.gov/health-information/disorders/muscular-dystrophy>

This page has information on muscular dystrophy including treatment, prognosis, research, and links to other resources.

<https://catalog.ninds.nih.gov/pubstatic//13-77/13-77.pdf>

Muscular Dystrophy: Hope Through Research booklet

6 National Paralysis Resource Center | Updated: 6/2025

Parent Project Muscular Dystrophy (PPMD)

<http://www.parentprojectmd.org>

1012 14th St., NW, Suite 500

Washington, D.C. 20005

Phone: 201-250-8440, 800-714-5437(Toll-free)

Email: info@parentprojectmd.org

PPMD is the largest nonprofit organization in the U.S. focused entirely on Duchenne.

The organization takes a comprehensive approach by funding research, raising awareness, promoting advocacy, connecting the community, and broadening treatment Options.

The information contained in this message is presented for the purpose of educating and informing you about paralysis and its effects. Nothing contained in this message should be construed nor is intended to be used for medical diagnosis or treatment. It should not be used in place of the advice of your physician or other qualified health care provider. Should you have any health care related questions, please call or see your physician or other qualified health care provider promptly. Always consult with your physician or other qualified health care provider before embarking on a new treatment, diet or fitness program. You should never disregard medical advice or delay in seeking it because of something you have read in this message.

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