Muscular Dystrophy

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.

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.

Duchenne MD

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

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

Myotonic dystrophy 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 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.

The disease 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.

Is there any treatment for muscular dystrophy?

There is no specific treatment for any of the various forms of MD. Physical therapy is often practiced preventing painful muscle contractures. And / or certain prescribed drugs 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, as noted before. Finally, cardiac abnormalities may require a pacemaker.

Sources: National Institute of Neurological Disorders and Stroke, National Institutes of Health, Myotonic Dystrophy Foundation

Websites

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
2894 South Coast Highway, Unit 1
Laguna Beach, CA 92651
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.

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.

Medline Plus: Spinal Muscular Atrophy
http://www.nlm.nih.gov/medlineplus/spinalmuscularatrophy.html
This page has information on muscular dystrophy including diagnosis and symptoms.

Muscular Dystrophy Family Fund
http://www.mdff.org/
PO Box 776
Carmel, IN 46082
Phone: 317-615-9140
MDFF 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 Institute 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

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.

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