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 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?

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

Websites

Muscular Dystrophy Association
https://www.mda.org/
National Headquarters
1016 W. Jackson Blvd., #1073
Chicago, IL 60607
Phone: 800-572-1717
Email: ResourceCenter@mda.org
MDA is dedicated to curing muscular dystrophy, ALS and related diseases by funding worldwide research. The Association also provides comprehensive medical and support services, and professional and public health education. MDA has more than 200 offices across the U.S., sponsors some 200 hospital-affiliated clinics, and supports more than 330 research projects around the world.

Centers for Disease Control: Muscular Dystrophy
www.cdc.gov/ncbddd/duchenne
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.

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/
1641 3rd Ave., #28K
New York, NY 10016
Phone: 646-429-0981
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).

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.

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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