



# **MDA<sup>®</sup>**

## **Fact Sheet**

## What is MDA?

The Muscular Dystrophy Association is a voluntary national health agency — a dedicated partnership between scientists and concerned citizens aimed at conquering neuromuscular diseases that affect more than 1 million Americans. Your local MDA office, one of 200 nationwide, is available to serve anyone with one of the diseases in the Association's program. MDA today is one of the world's leading voluntary health agencies fostering research and medical care. Programs available through your local MDA office are funded almost entirely by individual private contributors and cooperating organizations.

## How did MDA get started?

As late as 1950, very little was being done to combat muscular dystrophy. In that year, a small group of adults with muscular dystrophy and parents of affected youngsters decided to do something about this, and MDA was born. This group firmly believed that there are no incurable diseases, only diseases for which no treatments have yet been found. That basic philosophy has animated MDA ever since.

## What diseases does MDA strive to defeat?

**Muscular dystrophies:** There is no single disease called muscular dystrophy. The term designates a group of hereditary neuromuscular disorders, which vary in inheritance pattern, age of onset, initial muscles attacked and rate of progression.

**Motor neuron diseases:** This is a group of progressive diseases chiefly characterized by degeneration of the motor nerve cells in the spinal cord. They include amyotrophic lateral sclerosis (ALS) and several forms of spinal muscular atrophy (SMA).

**Diseases of the neuromuscular junction:** Myasthenia gravis is an autoimmune disease characterized by fluctuating weakness resulting from a failure in the transmission of sig-

nals from nerves to muscles. The disease initially affects eye movement, facial expression, chewing, swallowing and respiration, and later can affect arm and leg muscles. Lambert-Eaton (myasthenic) syndrome is associated with initial weakness of the muscles of the shoulders and thighs. Congenital myasthenic syndromes result from genetic defects.

**Metabolic disease of muscle:** These progressive diseases affect voluntary muscles and are characterized by inherited chemical deficiencies — usually of a specific enzyme such as phosphorylase (McArdle disease), acid maltase (Pompe disease) or carnitine palmitoyl transferase.

**Diseases of peripheral nerves:** These diseases affect the peripheral nervous system, causing progressive weakness and atrophy of muscles. Friedreich's ataxia is characterized by shaky movements and unsteadiness due to degeneration of both peripheral nerves and certain nerve cells in the brain and spinal cord. Charcot-Marie-Tooth disease (CMT) is characterized by progressive weakness of the feet, lower legs, hands and forearms, and a mild loss of sensation in the limbs. It results from degeneration of peripheral nerves that control muscle contraction and sensation.

**Inflammatory myopathies:** This group of potentially debilitating diseases is characterized by progressive weakness and inflammation of skeletal muscle and is attributed to disturbances in the immune system. Polymyositis can cause weakness of the pelvis, shoulders and limbs, as well as difficulty in swallowing. Dermatomyositis can cause similar symptoms and is accompanied by a rash and other changes in the skin. Inclusion-body myositis causes atrophy of the arms and legs, usually in people over 50.

**Myopathies due to endocrine abnormalities:** Disorders of the thyroid gland can sometimes affect muscle function.

Hyperthyroid myopathy is characterized by muscle weakness and muscle wasting. Hypothyroid myopathy causes delayed muscle relaxation, stiffness and painful cramps.

**Other myopathies:** This is a group of unrelated genetic muscle disorders, usually named for peculiarities seen in biopsies of muscle tissue. Central core disease, usually apparent at birth, causes skeletal deformities and diffuse weakness. Nemaline myopathy, also usually apparent at birth, results in loss of muscle tone and progressive weakness in limb and trunk muscles. Myotubular myopathy causes poor muscle tone at birth and varying degrees of weakness in eye, facial, neck and limb muscles.

## What does MDA do?

MDA works in your community to combat neuromuscular diseases through (1) basic and applied scientific investigation, (2) local comprehensive programs of medical and support services, and (3) widespread professional and public health education. Thanks to the MDA Labor Day Telethon (brought to you and thousands of other viewers in your area by a nearby “Love Network” TV station), and to many other local fundraising projects involving all sectors of the community, MDA has been able to organize and maintain a variety of programs and services.

**Worldwide research:** Grants are awarded to physicians and scientists at hospitals, universities, nonprofit institutions and biotechnology companies in the United States and abroad who are seeking the causes of, and treatments and cures for, any of the disorders in MDA’s program.

MDA annually funds more than 300 research projects worldwide. Careful review by MDA’s medical, scientific and translational research advisory committees ensures resources are allocated to achieve maximum results.

MDA-supported scientists conducted the first U.S. trial of gene therapy for Duchenne mus-

cular dystrophy by injecting a miniaturized version of the gene for the muscle protein dystrophin into an arm muscle of boys with this disorder. This phase 1 safety study has paved the way for further gene therapy trials in this and other MDA-covered disorders. A trial of gene therapy for limb-girdle muscular dystrophy has yielded encouraging results.

MDA also is supporting development of compounds that allow muscle cells to skip defective parts of a gene and piece together the surrounding, correct parts. This strategy is designed to allow cells to produce functional protein molecules even when a genetic flaw is present.

Tests of an enzyme replacement therapy for the metabolic muscle disorder acid maltase deficiency (Pompe disease), derived in part from MDA-funded basic research, had lifesaving results for affected infants, and resulted in FDA approval in 2006 of the first definitive treatment for a genetic disease in MDA’s program.

In ALS (Lou Gehrig’s disease), MDA and the nonprofit ALS Therapy Development Institute (ALS TDI), of Cambridge, Mass., forged a historic partnership in January 2007 when they launched a \$36 million industrial-scale research-and-development program to identify potential ALS treatments — the largest drug discovery project to date in ALS. In 2010, MDA renewed its partnership with ALS TDI with a grant of \$2.5 million. MDA also is funding the development of a compound to block the synthesis of a toxic protein in familial ALS. MDA/ALS centers are the sites of dozens of trials of new and existing medications, and other treatments.

**Nationwide services:** MDA maintains the most comprehensive services program of any voluntary health agency, helping individuals and their families meet the challenges imposed by chronic, progressive diseases. Included among these services is a network of outpatient clinics; assistance with locating and repairing durable medical equipment;

support groups; youth summer camp programs; flu shots; and resource referrals.

**Network of clinics:** MDA clinics offer an interdisciplinary team approach toward initial diagnosis and follow-up care. Individuals affected by any of the diseases in MDA's program have access to a nationwide network of some 200 clinics staffed by top health professionals. These experts can advise about all aspects of medical management of a disease, including occupational, physical, speech and respiratory therapies. While all MDA clinics serve people with amyotrophic lateral sclerosis (ALS), MDA has designated some 40 facilities as MDA/ALS centers because of the amount of ALS research taking place there and the vast experience of the medical staff in dealing with this disease. To find your nearest MDA clinic, go to [mda.org](http://mda.org), or call (800) 572-1717.

**Summer camps:** MDA provides youngsters in your community with summer camping activities geared to their special needs. MDA annually sponsors nearly 80 weeklong camp sessions for some 3,500 campers. MDA camps are staffed by health professionals who volunteer their services. Campers are assisted by volunteer counselors who often continue their friendships with campers on a year-round basis.

Professional and public health education: MDA seeks to provide the power of information to those living with muscle diseases, and to increase knowledge and awareness of these diseases among scientists, physicians, nurses, therapists and the general public. The Association offers a wide array of educational materials, including the award-winning quarterly magazine *Quest*, and dozens of informational and scientific booklets, some in Spanish. These publications are available on MDA's comprehensive website at [mda.org](http://mda.org). In addition, MDA regularly convenes international scientific meetings about disease research and holds conferences of MDA clinic directors and their associates.

**Advocacy:** MDA's advocacy efforts are committed to making life better for people with muscular dystrophy and related muscle diseases by providing representation in matters of public policy and research advancement, nationally and internationally; and facilitating active involvement in these areas by the people it serves.

## MDA accreditation

MDA is the first nonprofit organization honored with the American Medical Association's Lifetime Achievement Award "for significant and lasting contributions to the health and welfare of humanity."

MDA has been designated a "Top-Rated Charity" by the American Institute of Philanthropy.

In your state, county and city, MDA operates in accordance with all ordinances and regulations governing fundraising activities, and has never been denied a license anywhere.

## Who supports MDA?

MDA's national sponsors include: Acosta Sales and Marketing Co.; Bally Total Fitness; Burger King; CITGO Petroleum Corp.; ClubCorp; Convenience Store Industry; Dr Pepper Snapple Group/7UP; ERA Real Estate; Harley-Davidson Motor Co.; International Association of Fire Fighters; Jiffy Lube; Lowe's Home Improvement; National Association of Letter Carriers; Safeway; 7-Eleven; and Tall Cedars of Lebanon of North America.

National youth organizations supporting MDA include: DECA; Kappa Alpha Order; The National Beta Club; and Universal Cheerleaders and Dance Associations.

## MDA's Purpose and Programs

The Muscular Dystrophy Association fights neuromuscular diseases through an unparalleled worldwide research effort. The following diseases are included in MDA's program:

### Muscular Dystrophies

Myotonic dystrophy (*Steinert disease*)  
Duchenne muscular dystrophy  
Becker muscular dystrophy  
Limb-girdle muscular dystrophy  
Facioscapulohumeral muscular dystrophy  
Congenital muscular dystrophy  
Oculopharyngeal muscular dystrophy  
Distal muscular dystrophy  
Emery-Dreifuss muscular dystrophy

### Motor Neuron Diseases

Amyotrophic lateral sclerosis (*ALS*)  
Infantile progressive spinal muscular atrophy  
(*Type 1, Werdnig-Hoffmann disease*)  
Intermediate spinal muscular atrophy  
(*Type 2*)  
Juvenile spinal muscular atrophy  
(*Type 3, Kugelberg-Welander disease*)  
Adult spinal muscular atrophy (*Type 4*)  
Spinal-bulbar muscular atrophy  
(*Kennedy disease*)

### Inflammatory Myopathies

Polymyositis  
Dermatomyositis  
Inclusion-body myositis

### Diseases of Neuromuscular Junction

Myasthenia gravis  
Lambert-Eaton (myasthenic) syndrome  
Congenital myasthenic syndromes

### Diseases of Peripheral Nerve

Charcot-Marie-Tooth disease  
Friedreich's ataxia  
Dejerine-Sottas disease

### Metabolic Diseases of Muscle

Phosphorylase deficiency (*McArdle disease*)  
Acid maltase deficiency (*Pompe disease*)  
Phosphofructokinase deficiency  
(*Tarui disease*)  
Debrancher enzyme deficiency  
(*Cori or Forbes disease*)  
Mitochondrial myopathy  
Carnitine deficiency  
Carnitine palmityl transferase deficiency  
Phosphoglycerate kinase deficiency  
Phosphoglycerate mutase deficiency  
Lactate dehydrogenase deficiency  
Myoadenylate deaminase deficiency

### Myopathies Due to Endocrine Abnormalities

Hyperthyroid myopathy  
Hypothyroid myopathy

### Other Myopathies

Myotonia congenita  
Paramyotonia congenita  
Central core disease  
Nemaline myopathy  
Myotubular myopathy  
Periodic paralysis



MDA's website, [mda.org](http://mda.org), is constantly updated with the latest research news and information about the diseases in its program. Follow MDA on Facebook, Twitter and YouTube.



[mda.org](http://mda.org) • [als.mda.org](http://als.mda.org) • (800) 572-1717

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