



Facts About Inflammatory Myopathies (Myositis)

MDA[®] Muscular
Dystrophy
Association
Fighting Muscle Disease

Dear Friends:

The first time I heard the word dermatomyositis (DM), I had to have it repeated and then spelled out for me.

I was scared and confused. I could barely comprehend what the doctor was saying to me. I was young, a newlywed, with a promising career as an opera singer, and had just been told I had a serious disease. I thought to myself, “What is happening to me?”

For months I had experienced trouble walking, climbing stairs, even singing. Now I learned that I had a rare inflammatory muscle disease that affects fewer than 20,000 people in the United States.

Perhaps you’re reading this booklet because you, or someone you love, also received a myositis diagnosis. As I did, you’re probably wondering what this will mean for your future, your family, your dreams. You may be feeling worried and confused — and even angry.

This booklet is designed to give you a clearer understanding of the causes, symptoms, complications and treatments of DM, polymyositis (PM) and inclusion-body myositis (IBM). You’ll learn that, although these inflammatory muscle diseases can cause great distress initially, with proper treatment the symptoms can be alleviated. In fact, it’s possible to recover partially or completely from PM and DM.

As soon as I received my diagnosis in 1996, I contacted the Muscular Dystrophy Association. My local MDA office was very helpful, providing information, answering all my questions and helping me get established at the MDA clinic, where a course of treatment was started.

It wasn’t easy, but slowly things got better and I felt my strength returning. At the beginning of my treatment, I couldn’t manage to climb even a couple of stairs. But a

few years later I walked up 120 steps to my hotel room when I sang in Rome. In 2000, I sang for a national audience on the Jerry Lewis MDA Telethon, and I continue to perform nationally and internationally.

I also continue to visit my MDA clinic, where my treatment is monitored and adjusted as needed. Local MDA staff also direct me to resources or simply let me know I’m not alone in coping with this disease. On page 11, you’ll find out more about the many MDA services available in your community.

It can be painful coming to terms with what life has handed you and making the necessary adaptations. I know it was for me. But like me, I hope you find that your myositis isn’t a dead end in your life’s journey.

I still face challenges due to DM, but I’ve learned these challenges can be successfully managed. I know support is there for me from my husband Troy, friends and family, my medical team, and even from laws such as the Americans with Disabilities Act. My singing career continues to grow and (because some of the drugs used to treat DM and PM make pregnancy inadvisable), my husband and I, through international adoption, are blessed with three children.

When I first received my DM diagnosis, it was important to me that I continue to pursue my dreams. DM hasn’t stopped me, but has motivated me to fight even harder to do what I love. I pray this also is true for you. And remember: As you face this challenge, you’re not alone in your fight!

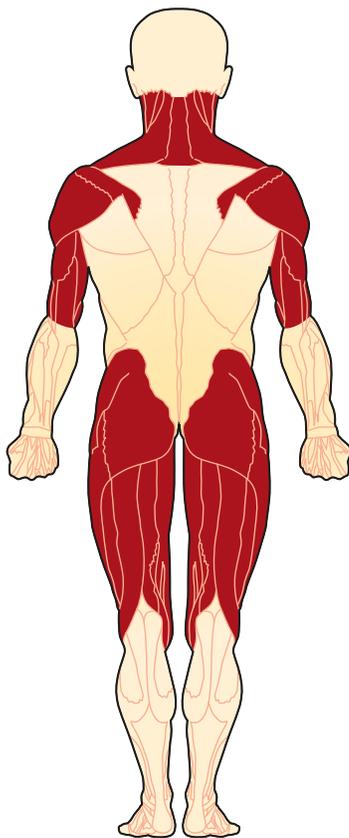
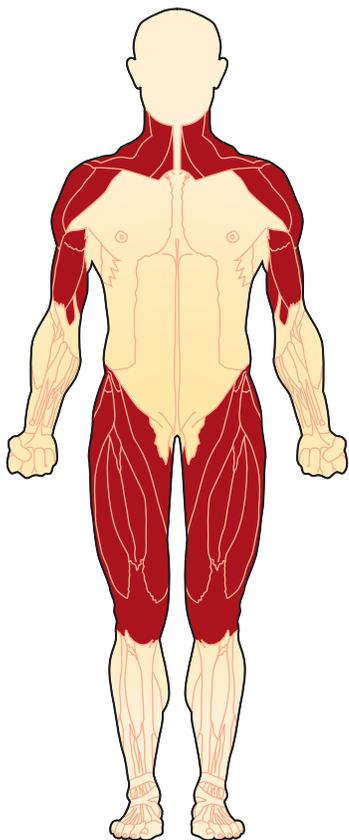
Sincerely,



Robin Chavez
Arlington, Texas



Robin Chavez
Photo: Christian Steiner



Polymyositis and dermatomyositis mostly affect the muscles of the hips and thighs, the upper arms, the top part of the back, the shoulder area and the neck.

What Are Inflammatory Myopathies?

The inflammatory myopathies are a group of muscle diseases that involve *inflammation* of the muscles or associated tissues, such as the blood vessels that supply the muscles. A *myopathy* is a muscle disease, and *inflammation* is a response to cell damage.

The inflammatory process leads to destruction of muscle tissue, and is accompanied by weakness and sometimes pain. Over time, there can be loss of muscle bulk (*atrophy*).

Normally, we think of inflammation, such as that following a sprained ankle or a dental procedure, as a condition that makes a part of the body hot, red and painful to touch. But inflammation also can be internal, causing tissue destruction in various organs. The common denominator in both types of inflammation is the presence of cells of the immune system in great numbers. Under a microscope, these can be seen “invading” the tissue as an army invades a city.

Another word for inflammatory myopathy is *myositis*. The *myo* root means muscle, and the *itis* root means inflammation; so a myositis is an inflammatory muscle disease.

Fortunately, for two of the three inflammatory myopathies in MDA’s program — *polymyositis (PM)* and *dermatomyositis (DM)* — effective treatments are available. New research is rapidly leading to increased understanding of these disorders and more successful treatments for them.

Although inflammatory myopathies can lead to great discomfort for at least a period of time, for the most part they aren’t life-threatening. In fact, many people recover partially or completely from PM and DM. The third inflammatory myopathy, *inclusion-body myositis (IBM)*, also isn’t life-threatening.

What causes inflammatory myopathies?

In most cases, the cause of an inflammatory myopathy is unclear. For some reason, the body’s immune system turns against its own muscles and damages muscle tissue in an *autoimmune* response.

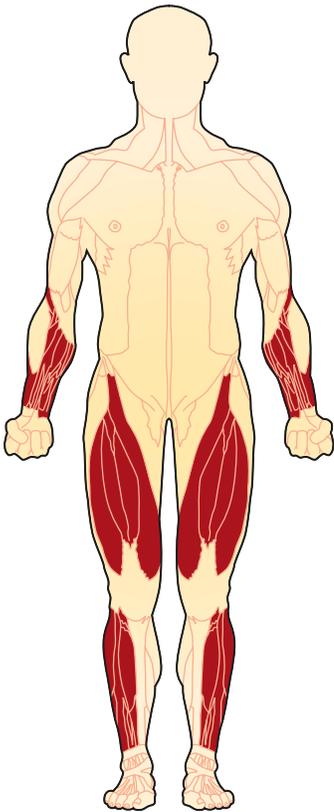
Viruses might be a trigger for autoimmune myositis. People with the HIV virus, which causes AIDS, can develop a myositis, as can people with a virus called HTLV-1. Some myositis cases have followed infection with the Coxsackie B virus.

There are reports of myositis following exposure to certain drugs. Among the drugs that have been suspected of contributing to myositis are carticaine (a local anesthetic), penicillamine (a drug used to lower copper levels in the body), interferon-alpha (mostly used to treat cancer and hepatitis), cimetidine (used to treat ulcers), carbimazole (to treat thyroid disease), phenytoin (used to treat seizures), and growth hormone. The vaccine for hepatitis B also has been implicated in some cases.

Recent research suggests that the mixing of blood cells of a mother and a fetus during pregnancy could lead to the later development of an autoimmune disease such as myositis in the mother or the child.

Inflammatory myopathies aren’t genetic disorders, although there may be genetic factors that make it more or less likely that an inflammatory myopathy will develop.

All these factors are being studied so that these diseases someday can be better understood, treated or perhaps prevented entirely. In the overwhelming majority of cases, there’s no clear cause for the development of myositis.



The first muscles affected in **inclusion-body myositis** are usually those of the wrists and fingers, and the muscles at the front of the thigh. The muscles that lift the front of the foot also may be affected.



DM can cause sensitivity of the eyes, and avoidance of sunlight during peak hours is recommended.

What are the forms of inflammatory myopathy?

There are three main types of inflammatory myopathy. These are:

- **polymyositis**, a disease in which the inflammatory cells of the immune system directly attack muscle fibers;
- **dermatomyositis**, a disease in which these cells attack the small blood vessels that supply muscles and skin;
- **inclusion-body myositis**, a disease of older people that appears to be partly inflammatory and partly a degenerative muscle disease.

People with polymyositis (PM) or dermatomyositis (DM) have a somewhat elevated risk of cancer. One theory about this is that, as the immune system tries to fight the cancer, it gets confused and attacks some of its own tissue. Adults may be asked to undergo testing for various types of cancer.

There's no apparent association of cancer with myositis in children, and inclusion-body myositis (IBM) isn't known to be associated with an increased cancer risk.

Can inflammatory myopathies be cured?

PM and DM are highly treatable diseases. Some people, especially children, recover completely from an inflammatory myopathy, while others experience greatly diminished symptoms for long periods of time. Several years of treatment to suppress the immune system may be necessary to achieve these results.

Those who don't recover completely may need to continue on at least a low dose of medication to control the autoimmune attack of PM or DM throughout their lives. Some permanent loss of strength and wasting of muscles sometimes occurs.

In other cases, the patient recovers his or her full strength and muscle size.

New findings on the genetic and environmental factors involved in autoimmune diseases should lead to more precise and effective drugs to treat them.

At the present time, there are no medications to treat IBM. Once acquired, it generally progresses slowly.

How are PM, DM and IBM diagnosed?

As with other muscle diseases, a doctor diagnoses an inflammatory myopathy by considering the patient's history, family medical history, and the results of a careful physical examination. This may be followed by some lab tests, perhaps of the electrical activity inside the muscles, and usually a muscle biopsy.

After a careful history and physical exam to document the pattern of weakness in the patient's muscles, a doctor who suspects myositis likely will order a blood test to check the level of *creatine kinase (CK)*, an enzyme that leaks out of muscle fibers when the fibers are being damaged. In PM and DM, the CK level is usually very high. In IBM, it may be only mildly elevated, or even normal.

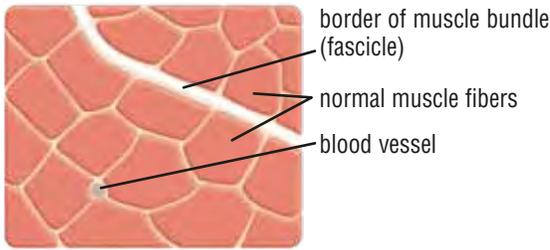
In some cases, the doctor may ask for a blood test for specific *antibodies*, proteins produced by the immune system in myositis and other autoimmune diseases.

Some of these antibodies appear to be specific to autoimmune muscle disease. One such antibody is called *Jo-1*.

The next step is sometimes an *electromyogram*, a test in which tiny needles are inserted into the muscles to test their electrical activity both at rest and when the person tries to contract the muscle. Inflammatory myopathies show a distinctive pattern of electrical activity that can

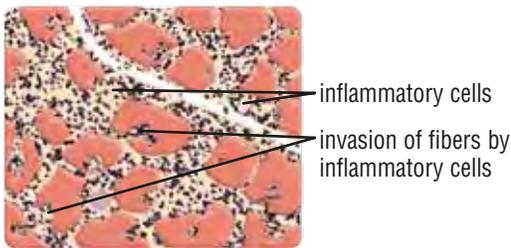
Microscopic Myositis

Normal Muscle



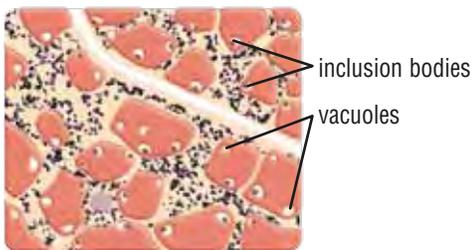
When normal muscle fibers are viewed under a microscope, they look like puzzle pieces that fit together neatly.

Polymyositis



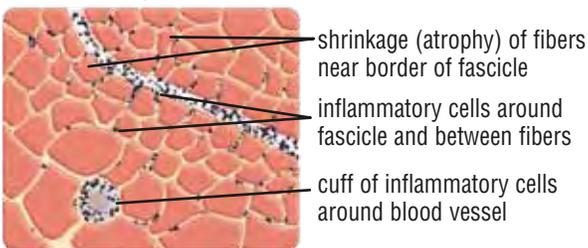
In polymyositis, inflammatory cells of the immune system invade previously healthy muscle cells, which become rounded and variable in size.

Inclusion-Body Myositis



Inclusion-body myositis is characterized by muscle fibers that contain empty, bubble-like spaces (vacuoles) and clumps of cellular material (inclusion bodies). Inflammatory cells can be seen between the fibers.

Dermatomyositis



In DM, inflammatory cells are concentrated around blood vessels at the borders of the muscle fiber bundles (fascicles), and fibers in this region often shrink. Inflammatory cells can sometimes be seen forming a cuff around blood vessels.

help differentiate them from other types of muscle disease.

A *nerve conduction velocity* test is sometimes performed. This test measures how fast a nerve impulse travels and how strong it is.

Sometimes these tests are used to rule out disorders that may mimic the symptoms of inflammatory myopathies.

A person with a suspected inflammatory myopathy is often asked to undergo a muscle biopsy, a procedure in which a small piece of muscle is removed for examination. This biopsy can enable the physician to pinpoint the diagnosis to a type of myositis (see “Microscopic Myositis,” left).

In PM, the biopsy generally shows the muscle fibers themselves being invaded by cells of the immune system.

In DM, the pattern of cellular invasion suggests that it’s the blood vessels in the muscles, and not the muscles themselves, that are the target of the attack. Muscle cells appear smaller than normal around the edges of bundles of muscle fibers, and capillaries are scarce in these regions.

The biopsy sample from a person with IBM is unique because of its *inclusion bodies*, for which the disease is named.

These “bodies,” which don’t appear in normal cells, contain clumps of discarded cellular material. Inflammatory cells can be seen invading muscle tissue, although some researchers

believe this invasion is secondary to the primary events in the muscle tissue, presumably those that cause the inclusion bodies to appear.

What happens to someone with polymyositis?

Although PM, DM and IBM have certain features in common, they differ in significant ways.

PM is more common in females than males and usually begins after age 20. Over a period of weeks or months, several muscles become weak and gradually get weaker. Most affected are the muscles of the hips and thighs, the upper arms, the top part of the back, the shoulder area and the muscles that move the neck.

Many people with PM have pain or tenderness in the affected areas. The person may have trouble extending the knee, stepping down or climbing stairs. Lifting things, fixing the hair or putting things on a high shelf may be difficult. It can be hard to raise the head off the bed when lying down.

Swallowing muscles can be affected as well, leading to poor intake of food and weight loss.

PM also can affect the heart muscle, causing a condition called *inflammatory cardiomyopathy*. The muscles involved in breathing may be affected and a few patients develop some inflammation of the lung tissues themselves, another respiratory complication.



Doctors test the strength of various muscles in determining which type of myositis a person has.



Medications usually are effective at controlling the symptoms of PM.

Of course, the heart, respiratory and swallowing problems are the most serious effects of PM and need close monitoring.

Treatment of PM

The first drug used in the treatment of PM is usually a *corticosteroid*, such as prednisone. The treatment may involve high-dose oral prednisone on a daily, every other day, or other schedule; or intermittent, short courses of intravenous corticosteroids. Sometimes, prednisone is stopped and then has to be restarted several times during the course of the disease.

Prednisone is usually very effective at bringing inflammation under control, restoring for the most part the person's strength, as well as swallowing, breathing and heart functions.

But prednisone has many side effects, including unwanted weight gain, redistribution of fat to the face and abdomen and away from the limbs, thinning of the skin, bone loss, cataracts and psychological problems. For this reason, if long-term treatment is necessary, most doctors (and patients) want to lower the dose of prednisone as quickly as possible. This can be accomplished by adding one or more other medications to suppress the damage being caused by the immune system.

These medications include azathioprine, methotrexate, cyclosporine, cyclophosphamide — all “traditional” immunosuppressants that have been used for many years; and some newer drugs, such as mycophenolate mofetil and tacrolimus. (See the chart on page 9 for more details about these medications.)

Although most people tolerate these medications without difficulty, they carry their own risks, such as flu-like symptoms, a lowered white blood cell count (which can predispose the patient to infection) and liver toxicity. Many are associated with an increased risk of cancer.

Some patients have responded well to intravenous infusion of antibodies culled from donors. This treatment — known as *intravenous immunoglobulins*, or *IVIg*, may seem strange in a disease that's probably caused by an immune response in the first place, but the extra antibodies seem to “confuse” the immune system and at least temporarily alleviate the attack on muscle.

Gently progressive physical therapy, such as that taken in a swimming pool, can be very helpful in maintaining strength. Range-of-motion exercise (putting a joint through its normal movement range), particularly of the shoulders, is helpful in keeping the joints supple.

Some people may need a cane, walker or even a wheelchair during acute flareups of PM.

Many people eventually recover much or all of their muscle strength and function, although they may relapse and lose function if they stop taking medications.

Plasmapheresis, a “blood-cleansing” process to remove antibodies, was at one time used in PM and DM, but is rarely used in these diseases today.

Immunosuppressant drugs and/or IVIg treatments are now considered more effective.

What happens to someone with dermatomyositis?

For many decades, DM was considered “polymyositis with a rash.” It's now known that the two diseases have some fundamental differences, but for most doctors, it's still the skin (“dermato”) manifestations of DM that make it a distinct disorder among the muscle diseases.

In DM, a distinctive reddish or purplish rash, presumably due to inflammation of surface blood vessels, may occur over the



A reddish or purplish rash and scaly, rough skin are typical in DM.



Because DM can affect the lung tissues, regular respiratory tests are recommended.



Some people with DM, when treated properly, recover completely.

face, neck and chest; on the shoulders and upper back, resembling a shawl; and/or on the elbows, knees and ankles. The eyelids may appear as if eye shadow has been applied.

The skin may be scaly, dry and rough. Sometimes it looks like a sunburn.

Unfortunately, the skin involvement in DM isn't limited to rashes.

A condition called *calcinosis*, in which calcium is deposited just under the skin in hard, painful nodules, also can occur, and seems to be more common in children with DM.

Inflammation of the fat lying just under the skin, called *panniculitis*, also can occur, causing tenderness and feeling like little bumps.

The muscles of the shoulders, upper arms, hips, thighs and neck display the most weakness. As in PM, the swallowing muscles can be involved, and a few people have difficulty chewing because of muscle weakness.

The weakness usually becomes noticeable over the course of several weeks, but it can move faster (days) or more slowly (months).

Joint pain with or without true arthritis (joint inflammation) can be part of DM.

Constriction of the blood vessels around the heart and inflammation of the heart muscle tissue can lead to cardiac complications.

Inflammation of the lung tissues also can occur, as in PM.

Patients with DM can have some inflammation of the blood vessels of the intestinal tract, eyes and kidneys, and these organs can be damaged as a result.

Treatment of DM

The treatment plan in DM is very similar to that used in PM, with drugs that suppress the immune system the mainstay of therapy.

Avoidance of sun exposure during peak sunshine periods and use of sunblock and protective clothing are recommended to avoid exacerbating the skin aspects of the disease.

In children, the disease usually begins between ages 5 and 14 and is more common in girls. It often announces itself with fatigue, fever and a rash, with the muscle pain and weakness following.

DM can be systemic, with many organs involved. Muscle weakness, gastrointestinal problems, joint inflammation and calcium deposits under the skin may last for a few years.

Children with DM are treated with the same medications and therapies as adults. They're more likely than adults to eventually recover completely.

Children may have to be kept out of physical education classes during periods of acute disease activity.

Myositis and You: A Guide to Juvenile Dermatomyositis for Patients, Families, and Healthcare Providers, is a 480-page book written by experts in this disease and the grandmother of a child affected by it. Published in 2007, it's available at bookstores and through the Internet.

What happens to someone with inclusion-body myositis?

Unlike PM and DM, IBM is a disease primarily of men instead of women, and mainly of those older than 50.

The disease also affects different muscles than the other inflammatory myopathies.



Many people with IBM remain able to walk but use wheelchairs for long distances.



Weakness caused by IBM may require modification of some activities. This man continues horseback riding but now uses a platform to mount the horse.

IBM usually begins with the gradual onset of slowly progressive weakness in the muscles of the wrists and fingers, and those at the front of the thigh (quadriceps). The muscles that lift the front of the foot also may be affected. The weakness may not be the same on both sides of the body.

Trouble with gripping a shopping bag or briefcase, and tripping, are common experiences. About a third of patients have some weakness of the swallowing muscles.

The heart and lung involvement seen in PM and DM is not part of the IBM picture.

IBM is generally a slowly progressive disease, and life expectancy isn't significantly affected. Most people with IBM remain able to walk, although they may require a cane or wheelchair for long distances. Some are more severely affected, becoming gradually more disabled and needing wheelchairs within 10 or 15 years of the first symptoms.

Treatment of IBM

Treatment with drugs that suppress the immune system has been tried in IBM, but in general hasn't been effective. Some physicians may try corticosteroids or other medications that alter the immune response if the patient wishes this treatment, but many feel that side effects outweigh any subtle benefits that might occur with these drugs in IBM.

There are some genetic forms of IBM in which, for the most part, inflammation isn't a major part of the picture. For this reason, these forms are often called *inclusion-body myopathy* (muscle disorder), leaving out the "itis" in the disease name to reflect the relative lack of inflammation.

Genetic inclusion-body myopathies can be inherited in either a *dominant* or a *recessive* pattern. Dominant genetic disorders require only one genetic flaw to

show themselves. Recessive disorders require that both parents pass on a flaw in the same gene before their offspring can show signs of the disease.

PM and DM Treatments

Medication or treatment	How it works	Comments
corticosteroids prednisone tablets (Deltasone); intravenous methylprednisolone sodium succinate (Solu-Medrol)	Dampens inflammation and immune response by interfering with processing of antigens and with early triggering of T-cell and B-cell production and later proliferation of B-cells and T-cells. These cells are produced by the immune system in autoimmune diseases such as PM and DM.	Can be taken orally as prednisone and related compounds; also available for intravenous use. Many side effects with long-term, high-dose therapy, such as weight gain and redistribution of fat to face, abdomen and upper back; thinning of skin; susceptibility to infection; bone loss; muscle damage; cataracts; elevated pressures in eyes (glaucoma); psychological disturbances; high blood pressure; high blood sugar; growth slowing in children.
azathioprine (Imuran)	Interferes with proliferation of B-cells and T-cells.	Can suppress production of several types of blood cells, so cell counts must be monitored; increases risk of cancer.
methotrexate (Rheumatrex, Folex, Mexate)	Interferes with proliferation of B-cells and T-cells.	Can cause liver damage; used in higher doses to treat cancer.
cyclosporine (Neoral, Sandimmune)	Keeps T-cells from stimulating production of more T-cells and B-cells (“upstream” of azathioprine and methotrexate action).	Doesn’t affect production of cells other than T-cells and B-cells; can cause kidney damage, infection, high blood pressure, tremor and excessive hair growth.
cyclophosphamide (Cytoxan)	Interferes with proliferation and activity of B-cells and T-cells.	Also used in cancer; toxic to many kinds of cells, including those of the blood and bladder; can cause sterility in both sexes.
mycophenolate mofetil (CellCept)	Interferes with proliferation of B-cells and T-cells.	Can cause diarrhea, vomiting, infection (particularly with cytomegalovirus); increases risk of cancer, especially lymphomas; causes depletion of certain blood cells.
tacrolimus (Prograf, old name FK506)	Keeps T-cells from stimulating production of more T-cells and B-cells (“upstream” of azathioprine and methotrexate action).	Can damage kidneys; can cause headaches, tremors and sleep difficulties; diarrhea, nausea and vomiting; high blood pressure, high blood sugar and high blood levels of potassium; increases risk of infection and lymphomas. Drug breakdown interfered with by grapefruit juice; potential for kidney damage increased by some anti-inflammatory drugs.
hydroxychloroquine sulfate (Plaquenil)	Mechanism not understood; used in arthritis, lupus, malaria; can be used to reduce steroid dosage in myositis, particularly in children.	Can treat muscle symptoms and dermatomyositis rash. Can cause damage to eyes’ retinas or corneas; regular eye exams needed.
infusion of mixed immunoglobulins; IVIg (Gammar, Gammagard, Sandoglobulin, others)	Has complex actions on immune system, such as providing antibodies against patient’s own antibodies; interfering with immune-system reaction to antibody-marked cells; interfering with blood-transported chemicals released by immune system; interfering with activation and maturation of T-cells and B-cells.	Doesn’t affect production of cells other than T-cells and B-cells; can cause kidney damage, infection, high blood pressure, tremor and excessive hair growth.
plasmapheresis	Removes antibodies and proteins made by the immune system from the blood and returns “cleansed” blood to patient.	Very rarely used in myositis since 1992 study showed it was no more effective than placebo; some think it’s useful when combined with immunosuppressant drugs.

MDA's Search for Treatments and Cures

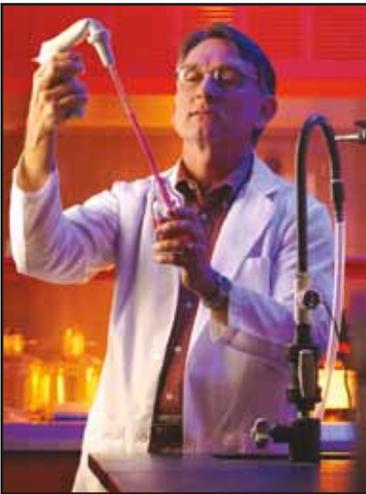
The MDA website is constantly updated with the latest information about the neuromuscular diseases in its program. See the latest research news at www.mda.org.

Researchers supported by MDA are studying the underlying mechanisms that cause inflammatory myopathies.

Several MDA projects are centered around understanding precisely what triggers the immune system to mistakenly attack muscle tissue, with the ultimate goal of stopping this type of attack.

MDA-supported researchers are studying the inflammatory myopathies that occur spontaneously in dogs to see if they offer insight into disease mechanisms, and the Association has funded a screen of approximately 110,000 candidate compounds for their ability to inhibit production of unwanted immune system cells.

In inclusion-body myositis, where some of the problem may not be autoimmune or inflammatory, researchers are working with mice with an IBM-like condition in an attempt to break up the abnormal clumps of cellular material in muscle fibers that characterize this disease.



MDA Is Here to Help You

The Muscular Dystrophy Association offers a vast array of services to help you and your family deal with DM, PM or IBM. The staff at your local MDA office is there to assist you in many ways. The Association's services include:

- nationwide network of clinics staffed by top neuromuscular disease specialists
- MDA summer camps for kids with neuromuscular diseases
- help with locating durable medical equipment through its national equipment program
- financial assistance with repairs or modifications to all types of durable medical equipment
- annual occupational, physical, respiratory or speech therapy consultations
- annual flu shots
- support groups for those affected, spouses, parents or other caregivers
- online support services through the e-community *myMDA* and through *myMuscleTeam*, a program that helps recruit and coordinate in-home help

MDA's public health education program helps you stay abreast of research news, medical findings and disability information through magazines, publications, educational speakers, seminars, videos and newsletters.

MDA's website at www.mda.org contains thousands of pages of valuable information, including disease specifics, research findings, clinical trials and past magazine articles.

Everyone registered with MDA automatically receives *Quest*, MDA's award-winning quarterly magazine. *Quest* publishes detailed articles about research findings, medical and day-to-day care, helpful products and devices, social and family issues, and much more. Other MDA publications can be found at www.mda.org/publications; many booklets are available in Spanish. Ask your local office for "MDA Services for the Individual, Family and Community" and for help with obtaining copies of other publications.

If you have any questions about DM, PM or IBM, someone at MDA will help you find the answer. To reach your local MDA office, call (800) 572-1717.



On the cover:
Giovanna was treated for polymyositis several years ago. After she recovered and stopped taking medication, some residual weakness in her hips and upper arms remained, but she's been able to walk without aids, swim and work as a restaurant hostess.



MDA's website, 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.



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P-199W 7/11

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