



Facts About Rare Muscular Dystrophies

Congenital (CMD), Distal (DD), Emery-Dreifuss (EDMD) & Oculopharyngeal Muscular Dystrophies (OPMD)

What is Muscular Dystrophy?

The muscular dystrophies are a group of genetic diseases that cause weakness and muscle wasting, primarily in the skeletal or voluntary muscles (those we control such as the muscles of the arms and legs). The four types of muscular dystrophy (MD) described in this pamphlet — *congenital muscular dystrophy (CMD)*, *distal muscular dystrophy (DD)*, *Emery-Dreifuss muscular dystrophy (EDMD)* and *oculopharyngeal muscular dystrophy (OPMD)* — are among the rarer forms of muscular dystrophy. Because they're less common, they can be difficult to diagnose, and many questions remain to be answered about their symptoms and progression.

The congenital muscular dystrophies and the distal muscular dystrophies (sometimes called *distal myopathies*) are both groups of muscle disorders. Emery-Dreifuss and oculopharyngeal each appears to be a single form of muscular dystrophy in terms of symptoms (although EDMD and OPMD can have more than one genetic cause).

Most people with muscular dystrophy experience some degree of muscle weakness during their lifetimes, but each of the four disorders described in this pamphlet affects different muscle groups and may have different accompanying symptoms. Because muscle weakness usually progresses over time in the muscular dystrophies, lifestyle changes, assistive devices and occupational therapy may be needed to help a person adapt to new situations.

What causes Muscular Dystrophy?

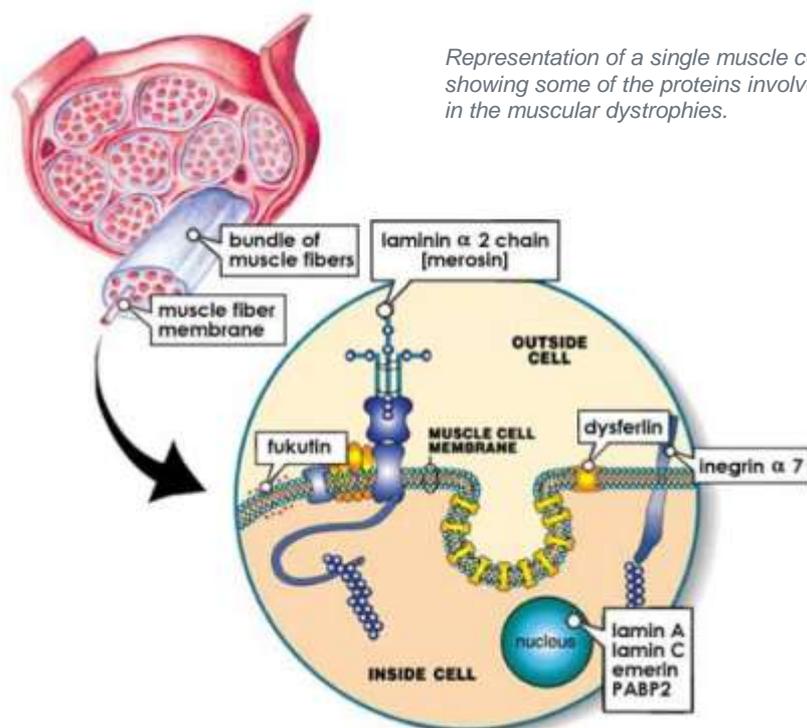
All the forms of muscular dystrophy are inherited — that is, they're caused by *mutations* (mistakes) in a person's *genes*. Our genes are made of DNA and they reside in our chromosomes. Each gene contains the "recipe" for a different protein and its variations, and these proteins are necessary for our bodies to function correctly.

When a gene has a mutation, it may make a defective protein or none at all. Most commonly, missing or defective proteins in the muscles prevent muscle cells from working properly, leading to symptoms of muscular dystrophy, including muscle weakness and wasting over time.

Muscles are made up of bundles of fibres (cells) (see diagram below). Groups of proteins along the membrane surrounding each fibre and within the cell help to keep muscle cells working properly. When one of these proteins is absent or inadequate (because a gene fails to make it properly), the result can be a form of muscular dystrophy. Absence of or defects in different proteins are among the causes of different types of muscular dystrophy. (The absence of other vital muscle proteins (not shown) leads to muscular dystrophies not covered in this booklet, such as Duchenne MD.)

Various forms of **congenital muscular dystrophy** arise from defects in proteins in or outside the membrane of the muscle cell (fukutin, integrin), or in the extracellular matrix, which attaches to the membrane (merosin or laminin-alpha-2.) Another membrane protein, dysferlin, is involved in **distal muscular dystrophy**.

The absence of some protein functions in the cell's nucleus leads to **Emery-Dreifuss muscular dystrophy** (emerin, lamin A, lamin C) or **oculopharyngeal muscular dystrophy** (PABP2).



What happens to someone with Muscular Dystrophy?

Most forms of muscular dystrophy are *progressive* and they tend to worsen with time. However, age of onset and rate of progression can vary widely from one disorder to the next. Some, but not all, of these disorders can affect life expectancy. In many cases, advancing knowledge allows for treatment of the symptoms that are most likely to decrease life expectancy.

In most cases of muscular dystrophy, muscle mass in the affected regions may become visibly wasted (decreased in size), and the arms, legs or trunk may become so weak they eventually can't move. Some forms of muscular dystrophy are accompanied by *contractures*, or stiff joints, and some are accompanied by *scoliosis*, or spinal curvature.



Forms of muscular dystrophy that affect the muscles used for *swallowing* may require that precautions be taken when eating or drinking so that food isn't aspirated into the lungs. Although most muscular dystrophies don't affect the *brain*, some are accompanied by brain changes that cause learning disabilities that range from slight to severe.

Finally, some forms of muscular dystrophy also affect the *heart*, and special precautions must be taken to monitor heart function. Each disorder has its own special areas of concern.

What Can Be Done to Treat Muscular Dystrophy?

There are currently no cures for any form of muscular dystrophy, but there are many *therapies* designed to help deal with common symptoms of the disease. For instance, contractures may be helped by physical therapy and sometimes tendon-release surgery, while scoliosis may respond to bracing or surgery, and heart problems may respond to medication or an implanted pacemaker. (Physical and occupational therapies can be arranged through your neuromuscular clinic.)

Many people with these forms of muscular dystrophy live very full lives. The team at your neuromuscular clinic will help you plan the best strategy for coping with your or your child's specific needs.



Does it run in the family?

On being told they have a genetic disorder such as muscular dystrophy, bewildered patients often ask, "But it doesn't run in the family, so how could it be genetic?" Muscular dystrophy *can* run in a family, even if only one person in the biological family has it. This is because of the ways in which genetic diseases are inherited.

Each form of muscular dystrophy follows one of three patterns of inheritance: recessive, dominant or X-linked. In brief, if a disease is *recessive*, two copies of the defective gene (one from each parent) are required to produce the disease. Each parent would be a *carrier* of the gene mistake (mutation), but wouldn't usually have the disease.

If a disease is *dominant*, then only one copy of the genetic defect is needed to cause the disease. Anyone with the gene mutation will have disease symptoms and can pass the disorder to children. If a disease is *X-linked*, it's passed from mother to son, while daughters can be carriers but don't generally get the disease.

Many times MD appears to have occurred "out of the blue," but in reality, one or both parents may be carriers, silently harboring the genetic *mutation* (a mistake in the gene). Many parents have no idea they're carriers of a disease until they have a child who has the disease. In rare cases, muscular dystrophy actually can occur "out of the blue" when a new mutation appears with a baby's conception, though neither parent carries the gene flaw. These are called *spontaneous mutations*, and, after they occur, they can be passed on to the next generation, thereby introducing the gene for a specific MD into the family.

The risk of passing on a form of muscular dystrophy to your children depends on many circumstances, including exactly which type of MD has been diagnosed. A good way to find out more about these risks is to talk to your doctor or genetic counsellor. You can also see our fact sheet "Genetics and Neuromuscular Diseases."



JERRY LEWIS
International Patron

Congenital Muscular Dystrophy

The term *congenital muscular dystrophy* (CMD) is actually the name for a group of muscular dystrophies that are united by the fact that muscle weakness begins in infancy or very early childhood (typically before age 2). Congenital diseases are those in which the symptoms are present at or soon after birth.

A diagnosis of CMD can be confusing because for many years the term was used as a "catchall" name to describe conditions that looked like other muscular dystrophies, but started much earlier or followed different patterns of inheritance. In recent years doctors have agreed that there are several different categories of "true" CMD, caused by specific gene mutations, and they're distinct from other muscular dystrophies. It's possible that some people who received diagnoses of CMD many years ago may actually have some other known form of muscular dystrophy with an unusually early onset.

Although children with CMD can have different associated symptoms, degrees of severity and rates of progression, most exhibit some progressive muscle weakness. This weakness, usually first identified as *hypotonia*, or lack of muscle tone, can make an infant seem "floppy." Later, infants and toddlers may be slow to meet motor milestones such as rolling over, sitting up or walking, or may not meet some milestones at all.

Some of the rarer forms of CMD are also accompanied by significant *learning disabilities*, or mental retardation.

What causes CMD?

The CMDs are caused by genetic defects that affect important muscle proteins. Most forms of CMD are inherited in an autosomal recessive pattern, but at least one form appears to follow a dominant pattern of inheritance. (For more about inheritance patterns, see "Does It Run in the Family?")

It isn't known why the CMDs cause muscle weakness earlier than other types of muscular dystrophy. One possibility is that the muscle proteins affected in CMD are required early in the development of an infant's muscle, while muscle proteins linked to other muscular dystrophies don't become important until the muscles begin to get a lot of use as a child grows.

It's important to note that just because the muscle weakness in CMD starts earlier, CMD isn't automatically more severe than other forms of muscular dystrophy. The degree and rate of progression of muscle weakness varies with different forms of CMD and from one child to the next.



A boy with congenital MD



What are the Types of CMD?

CMD isn't just one condition. Recent research shows that CMD can be divided into three major groups:

(1) *merosin-negative*, (2) *merosin-positive* and (3) *neuronal migration disorders*.

Merosin is a protein found in the thin layer of connective tissue that surrounds and supports each muscle fibre. People who have CMD either with or without merosin abnormalities have varying degrees of muscle weakness, and in some cases there's an accompanying learning disability. In neuronal (nerve cell) migration disorders, people have severe mental retardation and neurological disease that overshadows the muscle disorder.

(1) *Merosin-Negative CMD*

Children with merosin-negative CMD lack all or part of the muscle protein *merosin* (also known as *laminin alpha 2*). The degree of muscle weakness can range from severe (never walking) to mild (walking at 2 to 3 years), and depends on how much merosin protein a child is making.

This form of CMD progresses very slowly or, in some cases, not at all. Special problems include contractures, difficulty breathing and seizures (in 20 percent of cases). Intelligence is usually normal, but learning disabilities have been documented.

A distinctive diagnostic feature of this type of CMD is found by *magnetic resonance imaging (MRI)*. These brain images show changes in the white matter, which consists of nerve fibres that carry messages from the brain to the spinal cord. Despite the appearance on the MRI, those with merosin-negative CMD have few signs of brain impairment in everyday life.

(2) *Merosin-Positive CMD*

In this group of CMDs, muscle biopsies show the presence of merosin. But other proteins are missing in some forms of the disease.

Pure CMD

The clinical picture of pure CMD is similar to that of merosin-negative CMD. Muscle weakness may be mild to moderate, and complications may include contractures of the extremities. The brain appears normal and intelligence is normal.

CMD with Rigid Spine Syndrome (RSMD1)

This form of CMD is characterized by onset before 1 year of age with prominent neck weakness and poor head control. After some initial improvement, children gradually develop (by 3 to 7 years) stiffness or rigidity of the spine. To a lesser extent contractures of limb muscles are seen.

By the teens, the muscles that operate the lungs are affected, while limb muscle strength is less affected. Intellectual function is normal.



CMD can cause contractures in the wrists, ankles and other joints.



The defective gene leading to RSMD1 is on chromosome 1 and makes a protein called *SEPN1* or *selenoprotein*. The function of this protein is being investigated, but some studies suggest it protects the muscle against damage by free radicals.

Ullrich's Disease

Clinical characteristics of Ullrich's disease include hypotonia (loss of muscle tone) and multiple joint contractures at birth, with rigidity of the spine and marked *distal hyperlaxity* (loose joints) of hands and feet. The course is slowly progressive, causing muscle weakness and wasting. Intelligence is normal. Most people with Ullrich's disease have severe respiratory failure in the first decade of life.

Ullrich's disease may be inherited as an autosomal recessive or autosomal dominant disorder. The gene that, when defective, causes this disease produces a protein called *collagen* that also helps support the muscle fibre.

CMD with Integrin Deficiency

This recessively inherited form of CMD is caused by a gene responsible for producing a protein called *integrin alpha7*. This protein interacts with merosin and also surrounds and supports each muscle fibre. Children with integrin deficiency have hypotonia and weakness early in infancy associated with delayed milestones. Children usually don't walk until age 2 to 3.

(3) CMDs With Neuronal Migration Disorders

Fukuyama CMD (FCMD)

Fukuyama congenital muscular dystrophy is seen almost exclusively in those of Japanese descent. The disorder has been linked to a defect in a gene called *fukutin*, and the most common mutation is thought to have arisen in a single Japanese ancestor many years ago.

Muscle weakness in FCMD ranges from severe to mild, and people with the mildest cases are able to walk with assistance. Extensive brain abnormalities are usually accompanied by severe mental retardation, epilepsy, visual loss, and reduced life expectancy (about 11 to 16 years of age).

Muscle-Eye-Brain Disease (MEB)

This very rare form of CMD first described in Finland shares features with FCMD. Generally it's milder with survival ranging from early childhood to the seventh decade. It's accompanied by delayed motor milestones, severe mental retardation and vision problems. Mutations in the *POMGNT1* gene result in this disorder.

Walker-Warburg Syndrome (WWS)

This very rare form of CMD is similar to but more severe than MEB. People with this disorder have hypotonia and seizures. Severe mental retardation and multiple vision problems are encountered. The disorder is usually lethal in infancy. WWS can result from mutations in either the *POMT1* or *POMT2* genes.



Problems and solutions in Congenital Muscular Dystrophies

Contractures: Stiff or "frozen" joints (contractures) can develop as muscles weaken, but regular physical therapy designed to maintain range of motion at the joints can help combat this problem.

Scoliosis: Weak trunk muscles can lead to curvature of the spine, or scoliosis, which, in turn, can limit mobility and interfere with breathing. Regular physical therapy can help slow or prevent scoliosis, but corrective surgery may eventually be required.

Muscle weakness: Leg braces or a wheelchair may eventually be needed to help with mobility. An occupational therapist can help those with CMD find the best ways to perform day-to-day functions, often through use of assistive devices.

Respiratory insufficiency: Symptoms of respiratory insufficiency include morning headaches, fatigue, sleeplessness, weakened or softened voice, and unproductive cough. Advanced or severe weakness of the respiratory muscles (the diaphragm and rib cage muscles) may interfere with breathing. There are many options available to help with this problem, ranging from noninvasive nighttime ventilation to a tracheostomy.

Learning disabilities: Some children with CMD may have significant learning disabilities or mental retardation. Special education programs, begun as early as possible, can help a child maximize learning potential.

Seizures and vision problems: Specialists can address these problems with a variety of therapies.



Physical therapy is important in maintaining range of motion and reducing contractures.

Distal Muscular Dystrophy

First described in 1902, distal muscular dystrophy (DD), or distal myopathy, is the name of a group of disorders that primarily affect distal muscles (those farthest away from the hips and shoulders such as muscles in the hands, feet, lower arms or lower legs). Although muscle weakness is usually first detected in the distal muscles, with time, other muscle groups may become affected as well. Intellect isn't affected in these diseases.

What causes DD?

The DDs are caused by many different genetic defects, not all of which are yet known. Also, some of the DDs have been given different names based on various symptoms but may actually be caused by defects in the same gene.

Your own form of DD may or may not fit into one of these categories. Many of these diseases can vary from one person to the next and, in some cases, researchers are still in the process of sorting out what symptoms are linked to a particular genetic defect.



What are the types of Distal Muscular Dystrophy?

Welander's distal myopathy

This form of distal muscular dystrophy follows a dominant pattern of inheritance and usually has an onset between 40 and 50 years of age. Upper extremities tend to be affected first, then lower ones. The degree of muscle weakness involved can range from benign to severe. The cause of this disorder is not yet known although it has been linked to a region on chromosome 2.

Classification of Distal Muscular Dystrophies (DDs)		
Disease	Chromosome/Gene	Inheritance Pattern
Welander's distal myopathy	chromosome 2	dominant
Finnish/Markesbery distal myopathy	chromosome 2	dominant
Miyoshi distal myopathy	dysferlin	recessive or sporadic
Nonaka distal myopathy	GNE gene	recessive or sporadic
Gower's distal myopathy	chromosome 14	dominant
Hereditary inclusion-body myositis (HIBM)	GNE gene	recessive
Hereditary inclusion-body myositis (HIBM)	Unknown	dominant
Distal myopathy with vocal cord and pharyngeal weakness	chromosome 5	dominant

Finnish/Markesbery distal myopathy

Markesbery muscular dystrophy follows a dominant pattern of inheritance with weakness starting after age 40 in the lower extremities and progressing slowly to the upper extremities and trunk muscles. Cardiac problems can be a feature.

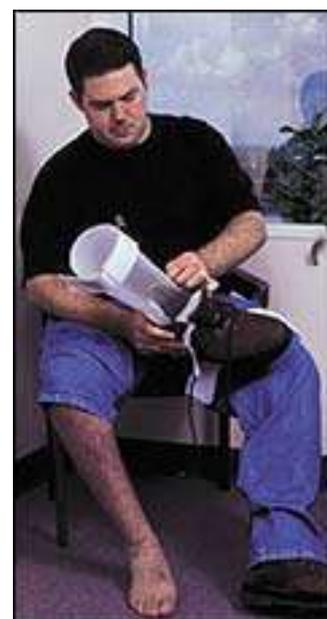
Finnish muscular dystrophy (also called *tibial MD*) can be severe or benign, and typically affects only people of Finnish descent. Those with only one defective gene experience mild weakness of the tibial leg muscles (front of the calf) sometime after age 40. Those with two defective genes have progressive weakness starting in childhood and may lose the ability to walk by age 30.

Finnish and Markesbery distal myopathies may be caused by defects in the same gene.

Miyoshi distal myopathy

This disorder is inherited in a recessive pattern and involves weakness that begins in the lower extremities, especially in the calf muscle. It can progress to other muscles as well. Symptoms usually begin between 15 and 30 years of age.

The genetic defect that causes Miyoshi myopathy has been mapped to the gene for a protein of unknown function called *dysferlin*. Defects in the dysferlin gene can also cause limb-girdle muscular dystrophy 2B, which causes muscle weakness in and



Orthoses, or leg braces, can support atrophied (wasted) lower leg muscles caused by distal MD.



around the hips and shoulders. People with the same genetic defect in their dysferlin genes can have either disease, and it isn't known what determines which pattern of symptoms a person gets.

Nonaka distal myopathy

Usually found in families of Japanese descent, this form of distal muscular dystrophy is inherited in a recessive pattern, and symptoms begin between ages 20 and 40. The anterior lower leg muscles (those in the front of the leg) are typically affected first, but the disease may progress to affect upper arm and leg muscles and neck muscles. The quadriceps muscles (in the thigh) tend to remain strong.

The disease is caused by a defect in the GNE gene, which is the same gene that causes recessive hereditary inclusion-body myositis.

Gower's distal myopathy

This disorder is inherited in a dominant fashion and has its onset from childhood to 25 years of age. Weakness is first seen in the leg and neck muscles and progresses slowly to include upper leg muscles, hands and more neck muscles.

Hereditary inclusion-body myositis (HIBM)

This disorder can be inherited either as a dominant or a recessive disease. The recessive form appears in the teens or 20s, and muscle weakness appears in both the distal muscles (those farthest away from the shoulders and hips) and in the proximal muscles (the shoulder and hip muscles).

The dominant form has its onset at 25 to 40 years, and weakness occurs in the distal and proximal limb muscles with slow progression. In both forms of HIBM the muscle tissue, as seen in thin cross sections, is characterized by the presence of tiny holes in the muscle fibres called *vacuoles*.

Distal myopathy with vocal cord and pharyngeal weakness

This disorder is inherited in a dominant pattern and has been linked to chromosome 5 in the same region as the gene that's defective in limb-girdle MD type 1A. Symptoms first appear between 35 and 60 years of age and include weakness of the hands, legs or voice. Difficulty in swallowing, *dysphagia*, may be a feature.

Problems and solutions in DD

Lower limb weakness: Weakness of the lower limb muscles may make walking or standing from a sitting position difficult. In some cases, a type of brace called an *orthosis* that's worn over the shoe and lower or upper leg can help with leg weakness. Eventually, a wheelchair may be needed.

Arm weakness: Your MDA clinic can refer you to an therapist who will help you get the most out of your arm muscles in performing day-to-day activities. Often, the therapist can recommend devices that may improve grip strength or help you elevate your arms to better perform activities such as brushing your teeth or hair.



An occupational therapist can show those with DD how to perform activities in spite of weak arm muscles.



JERRY LEWIS
International Patron

Emery-Dreifuss Muscular Dystrophy

Emery-Dreifuss muscular dystrophy is characterized by wasting and weakness of the muscles that make up the shoulders and upper arms and those of the calf muscles of the legs. Another prominent aspect of this disease is the appearance of contractures (stiff joints) in the elbows, neck and heels very early in the course of the disease. Finally, and very importantly, a type of heart problem called a *conduction block* is a common feature of EDMD and requires monitoring.

How is EDMD Inherited?

The most common form of EDMD is inherited in an X-linked pattern, but EDMD can also be inherited in a dominant fashion and, very rarely, in a recessive fashion. Although dominant, recessive and X-linked EDMD follow different patterns of inheritance, their symptoms are almost indistinguishable.

What Causes EDMD?

Researchers recently have identified the genes that, when defective, lead to the forms of EDMD. We now know that the gene that's defective in X-linked EDMD makes a small protein called *emerin*, which normally is located in the membrane that surrounds each cell's nucleus (the compartment in a cell's center that contains the chromosomes).

It isn't yet understood how the loss of emerin from the nuclear membrane in X-linked EDMD leads to the symptoms of muscular dystrophy. Some researchers think this lack of emerin interferes with the reorganization of the nuclear membrane after a cell has divided, leading to weak or dying cells.

Along these same lines, the gene that's been found defective in both the autosomal and recessive forms of EDMD contains the instructions for two closely related proteins called *lamin A* and *lamin C* that are also associated with the nuclear membrane of cells.

Again, it isn't yet known how the loss of lamins A and C leads to the symptoms of muscular dystrophy, but some research suggests that the nuclear membrane may become destabilized without the lamin proteins, leading to muscle breakdown.

Another question that remains to be answered is why the symptoms of EDMD are restricted primarily to the skeletal (voluntary) muscles and heart muscle, given that the emerin and lamin proteins are found in most tissues of the body.

What happens to someone with EDMD?

The symptoms of EDMD usually become apparent by 10 years of age, but the disorder tends to progress slowly. Early signs include "toe-walking" because of stiff Achilles' tendons in the heels, and difficulty bending the elbows. Later the signs of muscle weakness become more prominent but are still generally considered mild. Usually



EDMD leads to difficulty bending the elbows as well as muscle weakness.



cardiac problems are detectable by age 20, but they can occur at earlier stages in the disease as well. Intellect isn't affected.

The contractures that occur early in EDMD may make arm, neck, heel and spine movements difficult; however, the progression of muscle weakness seems to occur very slowly in EDMD and may not become a source of difficulty until later in life.

Cardiac problems can be life-threatening and may require the insertion of a pacemaker. Some women who are *genetic carriers* for X-linked EDMD may also be at risk for cardiac problems and this risk may increase with age (X-linked EDMD carriers don't tend to have muscle weakness or contractures).

Problems and solutions in EDMD

Contractures: Contractures develop early in EDMD and can worsen even if muscle strength doesn't change. Preventing contractures is difficult, but maintaining range of motion with physical therapy may help to slow their development. Surgical release of contractures is challenging because of their tendency to recur.

Cardiac conduction block: This form of heart problem occurs when the rhythm of the heartbeat is disrupted because the electrical impulses don't communicate properly between the heart's upper and lower chambers. Conduction block can lead to sudden cardiac arrest. By age 30, almost all those with EDMD will have some form of detectable cardiac involvement.

Fortunately, this problem is fairly easy to detect with an electrocardiogram, and the insertion of a pacemaker can be lifesaving.

In some cases, after pacemaker insertion, generalized chronic heart failure may eventually develop. There are many medications available to help combat chronic heart failure. *Anyone given a diagnosis of EDMD should be monitored regularly for signs of cardiac conduction block.*



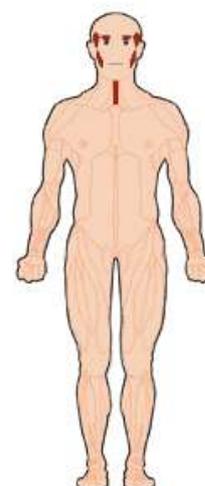
It's important for people with EDMD to monitor their heart function.

Oculopharyngeal Muscular Dystrophy

Oculopharyngeal muscular dystrophy (OPMD) is characterized by weakness of the muscles that control the eyelids (leading to droopy eyelids, a condition also known as *ptosis*), and by weakness of the facial muscles and pharyngeal muscles (those in the throat used for swallowing). It also affects limb muscles. Symptoms of the disease usually don't begin until the mid 40s or 50s, but can occur earlier.

OPMD is usually inherited as a dominant disease, but rare cases may show a recessive pattern of inheritance. When muscle tissue from a person with OPMD is examined under the microscope, clumps of proteins called *inclusions* are seen in the muscle cell nuclei (the cellular compartments that contain the chromosomes).

The disease is most common in French-Canadian families or families of French-Canadian descent. Research into the genealogy of these families has suggested that a single couple, Zacharie Cloutier and Sainte Dupont, who emigrated to Canada from





JERRY LEWIS
International Patron

France in 1634, may have harbored the genetic defect responsible for the majority of today's French-Canadian cases. OPMD can also affect people who aren't of French-Canadian background.

What causes OPMD?

The gene that's defective in OPMD was discovered in 1998, and is called the *poly(A) binding protein 2* or *PABP2* gene. Researchers suspect that in OPMD, the presence of extra amino acids in the protein made from a defective PABP2 gene causes the PABP2 protein to clump together in the muscle cell nuclei, perhaps interfering with cell function. The disease can often be diagnosed with a DNA test for the most common PABP2 mutation.

It isn't understood why symptoms of OPMD usually occur later in life, or why they're restricted to the specific muscle groups involved in OPMD.

What happens to someone with OPMD?

A person with OPMD may first notice drooping eyelids that gradually lead to tipping the head backwards to see properly. Alternatively, some people might first notice that they tend to choke frequently and may have other problems related to difficulty swallowing (called *dysphagia*). Most people eventually develop some degree of both ptosis and dysphagia.

Eventual weakness of the muscles in the face and limbs is common. For instance, many people with OPMD report problems with kneeling, bending, squatting, walking and climbing stairs. Double vision and a "breathy" quality of the voice may also occur.

Currently, there's no cure for OPMD, but many people with the disease find that treating symptoms as they occur is beneficial.

Problems and solutions in OPMD

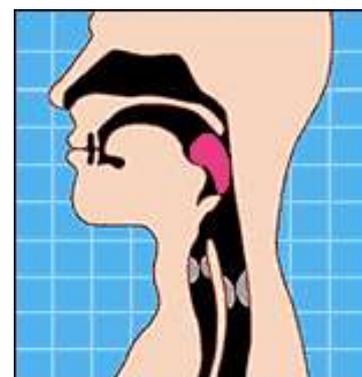
Dysphagia: Difficulty swallowing, or dysphagia, can cause a person to aspirate food or liquid into the lungs, which in turn may lead to a serious problem called *aspiration pneumonia*. If you find that you're choking frequently while eating or drinking, you may need to have your swallowing abilities evaluated by a professional. There are a number of techniques that may help treat dysphagia, ranging from holding the head in different positions to using commercial thickeners to give liquids a manageable consistency. In advanced cases, a nonsurgical procedure called *throat stretching* or a surgical procedure called a *cricopharyngeal myotomy* may be warranted. Tube feeding is another option for advanced cases.

Your neuromuscular clinic will refer you to a *speech pathologist* or an ear, nose and throat doctor as needed.

Ptosis: Droopy eyelids, or ptosis, can significantly impair vision and may lead to social awkwardness. This problem can be resolved with a type of eyelid reduction surgery called a *frontalis sling* performed by an oculoplastic surgeon.



Droopy eyelids are a common characteristic of OPMD.



OPMD may cause weakness in the muscles used for swallowing, which can lead to frequent choking.



Limb weakness: Trouble with picking up the feet when walking can lead to stumbling and falls. Leg braces, a cane or walker can help. Eventually, those with OPMD may need to use a wheelchair to make mobility more convenient.

Upper arm and shoulder weakness that limits function can be addressed with adaptive techniques through occupational therapy.

What tests are used to diagnose Muscular Dystrophy?

In diagnosing any form of muscular dystrophy, a doctor usually begins by taking a patient and family history and performing a physical examination. Much can be learned from these, including the pattern of weakness. The history and physical go a long way toward making the diagnosis, even before any complicated diagnostic tests are done.

The doctor also wants to determine whether the patient's weakness results from a problem in the muscles themselves or in the nerves that control them. Problems with muscle-controlling nerves, or *motor nerves*, originating in the spinal cord and reaching out to all the muscles, can cause weakness that looks like a muscle problem but really isn't.

Usually, the origin of the weakness can be pinpointed by a physical exam. Occasionally, special testing called *electromyography (EMG)* is done. In this kind of test, electricity and very fine pins are used to stimulate the muscles or nerves individually to see where the problem lies. Electromyography is uncomfortable, but not usually very painful.

Early in the diagnostic process doctors often order a special blood test called a *CK level*. CK stands for *creatine kinase*, an enzyme that leaks out of damaged muscle. When elevated CK levels are found in a blood sample, it usually means muscle is being destroyed by some abnormal process, such as a muscular dystrophy or an inflammation. Therefore, a high CK level suggests that the muscles themselves are the likely cause of the weakness, but it doesn't tell exactly what the muscle disorder might be.

To determine which disorder is causing CK elevation, a doctor may order a muscle *biopsy*, the surgical removal of a small sample of muscle from the patient. By examining this sample, doctors can tell a great deal about what's actually happening inside the muscles. Modern techniques can use the biopsy to distinguish muscular dystrophies from infections, inflammatory disorders and other problems.

Other tests on the biopsy sample can provide information about which muscle proteins are present in the muscle cells, and whether they're present in the normal amounts and in the right locations. This can tell the doctor and patient what's wrong with the cells' proteins and provide likely candidates as to which genes are responsible for the problem. The correlation between missing proteins on the muscle biopsy and genetic flaws isn't perfect, however. A neuromuscular clinic physician can help you understand these results.

An *MR (magnetic resonance) scan* may also be ordered. These scans, which are painless, allow doctors to visualize what's going on inside weakening muscles.





JERRY LEWIS
International Patron

Genetic tests, using a blood sample, can analyze the person's genes for particular defects that cause the rare muscular dystrophies, but these tests often aren't necessary for diagnosis or for determining treatment.