

The information in this fact sheet was adapted by the MDNSW (01/07) from the MDA USA fact sheet (updated 08/05) with their kind permission.



Facts about Myotonic Muscular Dystrophy

What is Myotonic Muscular Dystrophy?

Myotonic muscular dystrophy (MMD) is a form of muscular dystrophy that affects muscles and many other organs in the body. Unlike some forms of muscular dystrophy, MMD often doesn't become a problem until adulthood and usually allows people to walk and be pretty independent throughout their lives.

The infant form of MMD is more severe. Unfortunately, it can occur in babies born to parents who have the adult form, even if they have very mild cases.

The word *myotonic* is the adjective for the word *myotonia*, an inability to relax muscles at will. In MMD, the myotonia is usually mild. In fact, many people attribute it to "stiffness" or think they have arthritis. If anything is noticeable, it's usually difficulty with one's grip, for example when using a tool or writing instrument.

Myotonia isn't a feature of any other form of muscular dystrophy (although it occurs in other kinds of muscle diseases, where it can be severe). When a person suspected of having muscular dystrophy has myotonia, the diagnosis is likely to be MMD.

The term *muscular dystrophy* means slowly progressive muscle degeneration, with increasing weakness and wasting (loss of bulk) of muscles. The weakness and wasting of muscles generally present much more of a problem to people with MMD than does the myotonia. However, they usually aren't as severe as in some other types of muscular dystrophy.

MMD symptoms sometimes begin at birth. Infants with this disorder, *congenital* MMD, have severe muscle weakness, including weakening of muscles that control breathing and swallowing. These problems can be life-threatening and need intensive care. Myotonia isn't part of the picture in infants with MMD. MMD symptoms can also begin in children past infancy but not yet adolescents, although this is unusual. Generally, the earlier MMD begins, the more severe the disease is.

Myotonic muscular dystrophy is often known simply as *myotonic dystrophy* and is occasionally called *Steinert's Disease*, after a doctor who originally described the disorder in 1909. It's also called *dystrophia myotonica*, a Latin name, and therefore often abbreviated "DM."

There is, however, a distinct difference between the type that affects newborn infants — congenital MMD — and the type that begins in adolescence or adulthood — adult-onset MMD.

What causes Myotonic Muscular Dystrophy?

Myotonic muscular dystrophy is caused when a portion of either of two genes is larger than it should be. See "Does It Run in the Family?" to find out what scientists understand about how these genetic flaws cause MMD.



Weakness and wasting of voluntary muscles in the face, neck, and lower arms and legs are common in myotonic muscular dystrophy. Muscles between the ribs and those of the diaphragm, which moves up and down to allow inhalation and exhalation of air, can also be weakened.

The *chromosome 19* form of the disease, now called *type 1 MMD (MMD1)*, is the most common, and most of this fact sheet describes that form.

Type 2 MMD (MMD2), arising from an abnormality on *chromosome 3*, is less common, generally less severe, but not as well understood as the chromosome 19 form. Most of the information in this fact sheet is derived from studies of people with type 1 MMD.

What happens in Adult-Onset MMD?

It's reassuring to know that when MMD begins in the teen years or during adulthood, it's often only a moderately disabling condition with very slow progression. As one doctor put it, "Some people with MMD go through much of their lives without troubling or being troubled by the medical profession."

There can be troubling symptoms, however, for many people. Although many different parts of the body can be affected by MMD, most people with the disease have only some of the following symptoms. Most of the problems can be lessened with medical treatment.

Limb muscles

Weakness of the voluntary muscles, such as those that control the arm and legs, is usually the most noticeable symptom for people with adult-onset MMD.

The *distal* muscles — those farthest from the centre of the body — are usually the first, and sometimes the only limb muscles affected. The forearms, hands, lower legs and feet are the parts of the body that have these distal muscles. Over time, these muscles get smaller, so the lower legs and arms may appear thinner than the upper legs and arms.

People with MMD often notice that their grip is weak and that they have trouble using their wrist or hand muscles. At the same time, the muscles that pick up the foot when walking weaken, so the foot flops down, leading to tripping and falling. This is called foot drop.

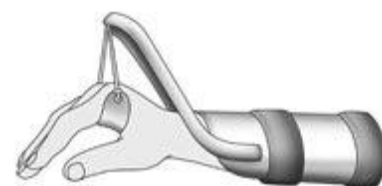
Some people can compensate for weak foot muscles by picking up the foot from the knee and walking with a "marching" step. Eventually, though, many people with MMD find that a cane or walker is helpful to compensate for foot and leg weakness.

A lower leg brace, called an ankle-foot orthosis or AFO, may be needed. A few people with MMD use a wheelchair or a power scooter for convenience when covering long distances.

Various devices that hold the hand in a good position for using a keyboard or writing or drawing can help compensate for weak wrist and hand muscles.

Head, neck and face muscles

The muscles of the neck, jaw and parts of the head and face may weaken, especially in MMD1. Weakness and loss of bulk in these muscles leads to a characteristic appearance doctors and experienced family members recognize as MMD. In men, early balding in the front part of the scalp is very common, adding to the distinct appearance of MMD.



A wrist support can hold the hand in a good keyboard, writing or drawing.



An ankle-foot orthosis (AFO) can keep the foot from flopping down and causing falls.

Eyelids may droop (called *ptosis*, but the “p” is silent), the temples appear hollow, and the face looks long and thin.

Severe ptosis can be troubling. It may be hard to hold the eyes open for reading, watching television or driving. Special glasses with “eyelid crutches” can hold the eyes open. You can’t buy these off the shelf, but a skilled optician can make them for you. Surgery can be done, but weakness often comes back, making it necessary to repeat the operation.

Weak neck muscles can make it hard to sit up quickly or lift one’s head straight up off a bed or couch. The stronger trunk muscles have to be used for these actions.

Muscle weakness generally has a somewhat different pattern in MMD2. Facial weakness is usually milder than in MMD1, while weakness of the upper part of the leg (thigh) occurs early in the disease. In MMD1, thigh weakness, if it occurs, comes late in the disease.

Breathing and Swallowing Muscles

Respiratory muscles can become weak in MMD, affecting lung function and depriving the body of needed oxygen. This is probably at least part of the reason many people with MMD feel sleepy much of the time.

The respiratory problems are further aggravated, many experts believe, by an abnormality in the brain’s breathing control centre. This abnormality can also lead to a condition known as *sleep apnoea*, in which people stop breathing for several seconds or even a minute many times a night while they’re sleeping.

A good way to treat respiratory muscle weakness is with a small, portable ventilator that pumps air into the lungs during the night. It’s usually used with a face mask that can easily be taken on and off. (This kind of breathing assistance can also be used during the day, but that’s usually not convenient or necessary.)

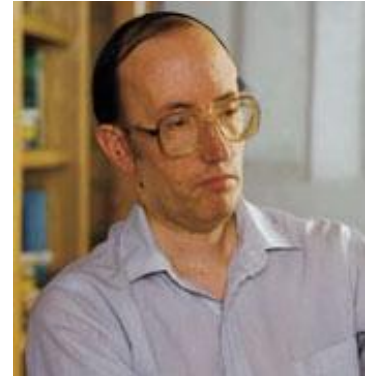
Devices and techniques to assist with coughing up secretions can be used, too, especially when the person with MMD has a cold or chest infection. Your doctor, or a respiratory specialist can help advise you about these devices and how to use them.

Swallowing muscles, if weakened, can lead to choking, or “swallowing the wrong way,” with food or liquid going down the trachea (windpipe) instead of the oesophagus (tube from the throat to the stomach). Swallowing is partly voluntary and partly involuntary, and both sets of muscles can be affected.

Vomiting can be very dangerous for a person with MMD whose swallowing muscles are weak. A head-down position is crucial to prevent inhaling the vomit — a possibly fatal problem.

A swallowing specialist can help people learn to swallow more safely and, if needed, how to change the consistencies of foods and liquids so they can be swallowed more easily. It’s important to watch for swallowing problems, such as a tendency to choke on food or drinks, and mention them to the doctor.

If swallowing difficulties are extreme (more common in congenital MMD than in adults with MMD), a feeding tube can be inserted into the stomach. It can later be removed if the problem resolves itself.



A long, thin face with hollow temples, drooping eyelids and, in men, balding in the front, is typical in myotonic dystrophy



The use of a portable ventilator with a face mask during the night can help compensate for weak breathing muscles and faulty breathing control by the brain.

Myotonia

The myotonia of voluntary muscles can make it hard for someone with MMD to relax the grip, especially in cold temperatures. Door handles, cups and tools may pose a problem, although many people never notice it.

Myotonia can affect other muscles, but usually it isn't noticeable. After sneezing, it can be hard to relax the muscles around the eyes. This can pose a driving hazard. If myotonia becomes troublesome, drugs can be used to treat it.

Heart Problems

The heart can be affected in adult MMD. Oddly, since MMD is mostly a muscle disease, it isn't the muscle part of the heart (which pumps blood) that's most affected, but rather the part that sets the rate and rhythm of the heartbeat — the heart's *conduction system*. It's common in MMD, especially after many years, to develop a *conduction block*, a block in the electricity-like signal that keeps the heart beating at a safe rate.

Fainting, near fainting or dizzy spells are the usual symptoms of conduction block, and these should never be ignored.

In the early stages, a partial conduction block may cause no symptoms but can be detected by an *electrocardiogram (ECG)*, a painless test of how the heart is beating.

Patients with MMD should be checked for *heart involvement* every 1-2 years. Heart involvement may occur in affected people who have very little muscle weakness. Usually your doctor will order both an ECG and an echocardiogram (ultrasound of the heart).

If the echocardiogram suggests that the heart is not contracting fully, your doctor may prescribe a medication such as an angiotensin converting enzyme inhibitor (ACE inhibitor – medications commonly used for treating high blood pressure and other heart disorders) to assist heart function. Conduction blocks can usually be corrected by a *cardiac pacemaker*, an electronic device that's surgically inserted near the heart to regulate the heartbeat.

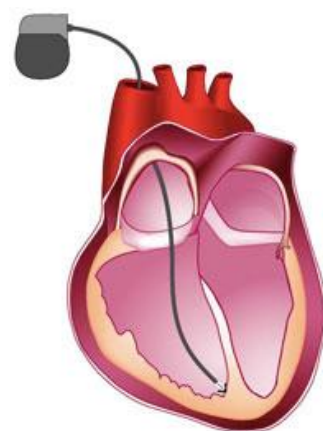
Not everyone with MMD needs treatment for heart problems, but everyone should be checked for them. Heart problems are thought to be less common in type 2 MMD than in type 1.

Internal Organs

Most of the internal organs in the body are hollow tubes (such as the intestines) or sacs (such as the stomach). The walls of these tubes and sacs contain involuntary muscles that squeeze the organs and move things (food, liquids, a baby during childbirth and so forth) through them.

In MMD, many of the involuntary muscles that surround the hollow organs can weaken and can also have myotonia. These include the muscles of the digestive tract, the uterus and the blood vessels.

Abnormal action of the upper digestive tract can impair swallowing. Once food is swallowed, the involuntary muscles of the oesophagus take over. These can have spasms and weakness, causing a feeling of food getting "stuck" and sometimes



A cardiac pacemaker can return the heartbeat to a normal rhythm.

leading to inhaling food into the lungs. Care in swallowing, sometimes with the advice of a specialist, may be needed.

The lower digestive tract — large intestine (colon), rectum and anus — can also be affected by weakness and spasm in MMD. Crampy pain, constipation and diarrhoea can occur. Your doctor can advise you about setting up a bowel routine and using diet and other treatments to help manage this kind of problem.

The gallbladder — a sac under the liver that squeezes bile into the intestines after meals — can weaken in MMD. People with MMD are probably more likely than the general population to develop gallstones. Symptoms are difficulty digesting fatty foods and pain in the upper right part of the abdomen. Surgery can be done if necessary.

Fortunately, most people don't have problems in urinating or holding onto urine in MMD.

Because of weakness and uncoordinated action of the muscle wall of the uterus, women with MMD often have complications in childbirth that can be serious for both mother and baby. These may involve excessive bleeding or ineffective labour. Sometimes a Caesarean section is advised, but surgery can also be a problem in MMD (see "Anaesthesia").

A pregnant woman with MMD has to be certain that all her doctors, including any who will manage the delivery, are well-informed about her neuromuscular condition. Disasters can result if this step is missing.

Blood pressure in MMD tends to be low. This is probably due to low tone of the smooth muscles in the blood vessels. It usually poses no problem and may even be one beneficial effect in MMD.

The Brain

Some people with type 1 MMD have been labelled by doctors and family members as slow, dull, uncaring, unenthusiastic or depressed. Only recently have researchers tried to get at the truth or untruth of these descriptions.

First, as with other aspects of MMD, there's a wide range in severity of the mental and emotional symptoms of the disorder. Some people function very well, others poorly, many somewhere in between.

Children born with the severe, congenital form of MMD1 have a lot of learning problems and may even be mentally retarded. They often need special education because of these disabilities.

In adults, severe mental impairment is less common, but an overall inability to "settle down to something," apply oneself to work or family life, concentrate or become engrossed in a task is often reported in MMD1. MMD2 doesn't seem to involve these sorts of problems, although there can be mild cognitive difficulties.

Adults with MMD1 often find they need much more sleep than do other people and may feel at the beginning of the day the way most people feel at the end of a long workday. This can be very hard for others to understand.

Recent research suggests that in MMD1 there may be abnormalities in the parts of the brain that determine the rhythm of sleeping and waking. Respiratory regulation and weakness of the respiratory muscles, along with irregular breathing during sleep, all combine to make this problem severe in some people (though not in everyone).

Weakness of the facial muscles, with drooping eyes, can add to an outsider's impression that the person with MMD1 is apathetic or dull. Facial expression can be misleading in this disorder. Facial weakness is mild in MMD2 and is less likely to confuse observers.

Daytime sleepiness can sometimes be helped with medication. One drug that can be used is *methylphenidate* (Ritalin). A newer drug is *modafinil* (Provigil). These drugs may work on the brain's sleep-wake cycle.

Another approach that can be tried is to coax the body into a better rhythm of sleeping and waking by going to bed and getting up at the same time every day no matter what the requirements of the day may be.

The Eyes

Cataracts — cloudy areas of the lens of the eye that can eventually interfere with vision — are extremely common in both types of MMD. Cataracts are caused by a chemical change in the lens, which gradually goes from clear to cloudy the way the clear part of an egg changes to white when cooked. Exactly why cataracts occur in MMD isn't known.

The person with a cataract may notice that things start to look blurry, hazy or dim, and that this worsens gradually over time. It often happens in both eyes but not necessarily at the same time or at the same rate.

Surgery can remove a lens that contains a cataract. Then, the surgeon either puts in an artificial lens, or the patient can wear special contact lenses or eyeglasses.

Vision correction with cataract surgery is quite good. However, with this operation or any procedure requiring anaesthesia, the medical team must be informed about the underlying MMD. Anaesthesia can pose special problems for the patient with MMD (see below).

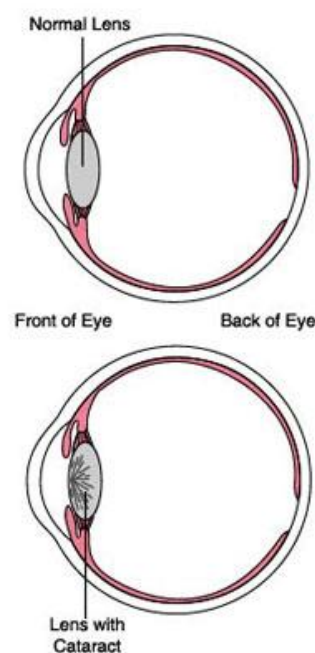
The muscles that move the eyes, as well as those that open and close them, can also be affected in MMD, and other eye problems can sometimes occur. Your primary care provider or clinician can refer you to an eye doctor (ophthalmologist) when eye problems need attention or for regular checkups.

Diabetes

If you read about MMD in books or on the Internet, you may find diabetes listed among the problems in this disorder.

Fortunately, most people with MMD don't have severe diabetes, but they may develop a mild type sometimes referred to as *insulin resistance* with high blood sugar. This means the body makes insulin (a hormone needed for the cells to take up and use sugars), but for some reason, the insulin isn't quite doing its job.

Your doctor may order blood and/or urine tests to see if you have insulin resistance or diabetes. If you do, you may be advised to change your diet or exercise habits or to



The eye's lens focuses light on the back part of the eye to allow vision. When cataracts cloud the lens, the visual image is no longer as clear.

take medication. Your doctor may refer you to a specialist or primary care physician for further treatment for diabetes.

High blood sugar affects a minority of people with MMD2.

Anaesthesia

An unusually high rate of complications and even deaths associated with general anaesthesia (given during any surgery) has been reported in people with MMD. This can occur even if the MMD is mild. In fact, these cases can be particularly dangerous, because the surgeon, anaesthetist and patient may be less likely to pay attention to the MMD when planning surgery.

Surgery can usually be safely undertaken with careful monitoring of cardiac and respiratory functions before, during and after the surgery. Be sure to tell the entire medical team, especially those responsible for the anaesthesia, that you or your family member has MMD. Have the anaesthetist and the neurologist communicate long before the surgery if at all possible.

What happens in congenital MMD?

The most serious form of MMD is the congenital (at birth) form of the disease. Congenital MMD has only been observed in MMD type 1. When a child with congenital MMD is born, it's almost always found that the mother has adult-onset MMD — even though her symptoms may be so mild that she doesn't even know she has the disorder.

Mothers with MMD can also pass on the adult-onset form. A child can also inherit the disease from the father, but it's almost always the adult-onset form. These unusual features aren't seen in other genetic disorders.

Weak Muscles

Babies with congenital MMD have very weak muscles and a lack of muscle tone (*hypotonia*). They appear floppy, have trouble breathing, and suck and swallow poorly.

In the past, many of these infants didn't survive. Today, with special care in neonatal intensive care units, such children have a much better chance of survival, but they enter childhood with multiple problems.

Respiratory support, such as artificial ventilation, will probably be needed, at least at first. Voluntary and involuntary aspects of respiration are likely to be affected in congenital MMD. Because swallowing muscles are affected, special feeding techniques or a feeding tube that goes into the stomach may be needed to provide adequate nutrition and prevent choking.

Children with congenital MMD have severe facial weakness, leading to a lack of facial expression and an upper lip that comes to a point — known as a *tented upper lip*.

Babies with congenital MMD are often born with *clubfeet* — a curvature of the feet and lower legs. Clubfeet need surgical correction for the child to be able to walk. The problem may be due to abnormal muscle development in the lower legs and feet during foetal life.



If you're planning surgery, be sure the neurologist, anaesthetist and surgeon know you have myotonic dystrophy.



A child born with congenital myotonic dystrophy is likely to have facial weakness and an upper lip that looks "tented." The eye muscles may also be affected.

Infants with MMD *don't* have myotonia. If they survive, however, they'll develop it later in life.

Intellectual disability

Infants born with congenital MMD are likely to have intellectual impairment, although this isn't always the case. This seems to be related to maldevelopment of parts of the brain, presumably caused by genetic abnormalities.

Some experts have suggested that the very high incidence of labour and delivery complications in mothers with MMD could also be a contributing factor to the learning difficulties seen in these babies. For this reason, it's very important to make doubly sure that everyone on the medical team is aware of and can work to minimize the risks surrounding labour and delivery to the mother and child with MMD.

Speech and Hearing Difficulties

The muscles involved in talking are often affected in congenital MMD. Hearing can also be impaired.

Therapy from a *speech-language pathologist* (in a medical centre) or *speech therapist* (in a school) can help. Even before a child enters school, early intervention programs are vital. Talk to your paediatrician, MD clinic physician or medical social worker about such programs.

Vision Problems

The eye muscles are affected and can cause the eyes not to work together; this condition is called *strabismus*. If severe, it can be corrected with surgery.

Cataracts, common in adult-onset MMD, *aren't* a feature of congenital MMD during early childhood. However, children with MMD are likely to develop them later.

Outgrowing Congenital MMD

Infants and children with MMD symptoms may "outgrow" many of the muscle-related aspects of the disorder as they mature. The mental retardation doesn't improve, but these children can learn if given the right tools and environment.

Unfortunately, despite early gains during childhood, all children with congenital MMD go on to develop the adult form of MMD when they reach adolescence or adulthood.

How is MMD Diagnosed?

Doctors who have experience with neuromuscular disorders find it easy to diagnose type 1 MMD. They can usually just look at a person, examine him and ask a few questions to make the diagnosis. Teen-agers and adults with MMD usually have a characteristic long face with hollow temples and, in men, early balding.

Many people tell the doctor about recurring abdominal pain, constipation or obstetrical complications. Others say their parents had some muscle problems.

Sometimes, an eye doctor will notice the particular type of cataract found in MMD and suspect the diagnosis, referring the patient to a neurologist.

Many people may not realize they have any trouble relaxing their grip, but others say they've had trouble letting go of a shovel, screwdriver or some other device, especially in cold weather.

The doctor may check for myotonia by lightly tapping the area just under the thumb with a rubber hammer. In most people, there is little or no response. In people with myotonia, there's a swift contraction of the muscle, which takes several seconds to relax.

The doctor may want to do electrical testing of the muscles and nerves, using an *electromyogram*, or *EMG*. In this exam, small needles are inserted into muscles to measure their electrical activity.

In a few cases, a *muscle biopsy* may be considered. In this test, a small piece of muscle is surgically removed for examination.

The doctor may move from the history and physical exam to a DNA test to confirm a diagnosis of MMD. The DNA test involves only a blood sample and, in almost all cases, can determine whether the family is affected by MMD. See 'Does it run in the family' below.

How is MMD treated?

At this time, there's no specific treatment that "gets at the root" of MMD. Treatment is aimed at managing symptoms and minimizing disability.

Canes, braces, walkers and scooters can help with mobility problems. Careful monitoring of cardiac and respiratory functions can lead to early treatment of these problems with cardiac medications, a cardiac pacemaker or assisted ventilation.

Medications and other treatments for constipation and other digestive tract complaints can be employed. Surgery for cataracts and either surgery or special eye crutches for drooping eyelids can markedly improve vision. New medications to treat excessive sleepiness can make life more enjoyable for the person with MMD and his or her family.

In children with the congenital form of MMD, early intervention is crucial. Hearing and vision abnormalities should be diagnosed and treated as soon as possible. Surgery for uncoordinated eye muscles and special education are among the interventions that can greatly influence a child's later success in life.

If you have a child with congenital MMD, it's very important to seek out an early intervention program through your muscle clinic, paediatrician, medical social worker, school system or other resources.



A cane can provide support when lower leg weakness makes walking hazardous.

Does it run in the family?

MMD is certainly a disease that runs in families. Both types are inherited in an *autosomal dominant* pattern, meaning it takes only one flawed gene to cause symptoms of the disease. So, if one parent has the disorder, every child of that person has a 50 percent chance of inheriting the gene that causes it.

If either the type 1 (chromosome 19) or the type 2 (chromosome 3) genetic abnormality is passed on, the child will almost certainly develop the disease. In MMD1, it will very often be more severe in the child than in the parent. In MMD2, increases in severity from generation to generation, if they occur, are smaller.

A 'Growing' Gene

In 1992, a landmark genetic discovery was made by three teams of scientists. They found in people with MMD an area of DNA (the basic genetic material that makes up our genes) on chromosome 19 that's larger than it should be.

The expanded DNA is in a gene that carries instructions for *myotonin protein kinase*. The expanded DNA isn't in the "working" part of the gene — the part that carries instructions for making protein. Instead, in MMD, the genetic flaw is in a part of a gene called the *untranslated* DNA, an area of DNA that the cell doesn't use for protein manufacturing. The experts were puzzled to find that an expanded section of this untranslated DNA could cause so much trouble, and the mystery still isn't entirely solved. It may be that this untranslated DNA interferes with normal gene processing.

There was more puzzlement to come. The expanded section of DNA was found to grow even more as it was passed from parent to child. This explained the long-observed phenomenon in MMD whereby children are generally more seriously affected by the disease than are their parents (called 'anticipation').

The expanding DNA also explains why children with the congenital form of type 1 MMD can be born to parents who have the less severe, adult-onset form. It doesn't, however, fully explain why this phenomenon occurs so often when mothers have MMD and so rarely when fathers do. It may have to do with a difference in the way egg cells, as opposed to sperm, are made in the body.

Then, in 2001, researchers identified a gene on chromosome 3 that carries instructions for a protein called *zinc finger 9*. When this gene contains an expanded section of DNA, it too causes MMD. That type of myotonic dystrophy, MMD2, is found chiefly in Northern Europeans or their descendants.

Today, scientists are investigating how the expanded areas of DNA cause the various symptoms of MMD. Ongoing research to answer these questions should lead to treatments for MMD.

Genetic Testing

Genetic testing for the expanded DNA that leads to either type of MMD can be performed. Prenatal testing is readily available for MMD1 and is offered in a few labs for MMD2.

People who have the MMD gene abnormality may have very few or even no signs of MMD, but they still have a risk of having children with the severe, congenital form of MMD. Therefore it is recommended that all people who are at risk of having inherited the MMD gene — those who have a parent, sibling or grandparent with MMD — should seek genetic counselling. Having a genetic test to confirm whether or not they have the expanded MMD gene may help them to make decisions about family planning.

A good way to find out more about the inheritance pattern in your family is to talk to your doctor or a genetic counsellor. For more information on how MMD is inherited, or how it can occur with no family history, see our fact sheet "Genetics and Neuromuscular Disease".

Search for treatments and cures

The years since the discovery of the genetic cause of MMD in 1992 have been fruitful ones for MMD research. Scientists are gaining understanding of how the expanded DNA section on chromosome 19 causes so many physiological changes. Such discoveries are likely to provide valuable insights for future treatment avenues.

In the meantime, scientists are also working to test drug treatments that may help symptoms in MMD. Among these are a drug that can make muscles more sensitive to insulin, one that may help improve muscle function and one that may relieve myotonia.

The ultimate “cure” for MMD would probably require finding a way to block the expanded area of DNA on chromosome 19 or chromosome 3 so that it would lose its toxic effect on cells. It’s not far-fetched to imagine that, in the future, this expanded section of DNA could be blocked or “silenced.”

Scientists around the world are studying the unusual biological mechanisms that underlie MMD and working on pathways to treatment.