MYOTONIC DYSTROPHY FOUNDATION

FAQS

What is myotonic dystrophy?

Myotonic dystrophy (DM) is a muti-systemic inherited disease that affects at least 1 in 8,000 people or 40,000 individuals in the US alone. Although often viewed as a muscle disease, individuals affected by DM may have skeletal muscle problems, heart function abnormalities, breathing difficulties, cataracts, issues with speech and swallowing (dysarthria and dysphagia), cognitive impairment, excessive daytime sleepiness, or diabetic symptoms. Any single individual is unlikely to have all or even most of these symptoms.

Myotonic dystrophy is one of the most variable and complicated disorders known. The systems affected, the severity of symptoms, and the age of onset of those symptoms vary greatly between individuals, even in the same family. In general, the younger an individual is when symptoms first appear, the more severe symptoms are likely to be. However, prognosis is as variable as the symptoms of this disease.

What are other names for myotonic dystrophy?

- Myotonic muscular dystrophy - often abbreviated as MMD
- Myotonia atrophica - a Latin name, not commonly used
- Dystrophia myotonica - a Latin name used by many doctors; often abbreviated as DM. The different types of DM are typically referred to as DM1 or DM2.
- DM1 is also known as Steinert's Disease, named for the German doctor who first identified this disorder in 1909.
- DM2 is also known as Proximal myotonic myopathy or PROMM.

Myotonic dystrophy should not be confused with other disorders with similar names (e.g.myotonia congenita [Thomsen's disease] and congenital muscular dystrophy).

What is the difference between myotonic dystrophy and muscular dystrophy?

Muscular dystrophy (MD) refers to a group of nine genetic diseases that cause progressive weakness and degeneration of muscles used during voluntary movement. Myotonic dystrophy (DM) is one of the muscular dystrophies. It is the most common form seen in adults and is suspected to be among the most common forms overall.

What are the types of myotonic dystrophy?

There are two well-defined types of the disease (DM1 and DM2) which have distinct but overlapping symptoms. Both DM1 and DM2 are characterized by muscle weakness and myotonia, heart abnormalities, cataracts and insulin resistance. In general, DM2 is less severe than DM1: fewer systems are affected, patients develop the disease only as adults, and the disorder's impact on everyday life is relatively less disruptive

In contrast, DM1 can occur from birth to old age. Symptoms vary greatly among patients, from minor muscle pain to serious respiratory and cardiac issues. The congenital form of DM1 is the most severe version and has distinct symptoms that can be life-threatening.

How do people get myotonic dystrophy?

Myotonic dystrophy is an inherited disease where a change, called a mutation, has occurred in a gene required for normal muscle function. The mutation prevents the gene from carrying out its function properly. The change is an autosomal dominant mutation, which means one copy of the altered gene is sufficient to cause the disorder. As a result, affected individuals have a 50% chance of passing on the mutated gene to their children. A child is equally likely to have inherited the mutated gene from either parent. If both parents do not have the disease, their children cannot inherit it. The congenital form of DM1 is inherited differently from the other types of myotonic dystrophy. Children with congenital myotonic dystrophy almost always inherit the disease from an affected mother.

How is myotonic dystrophy diagnosed?

A complete diagnostic evaluation, which includes family history, physical examination, and medical tests, is typically required for a presumptive diagnosis of myotonic dystrophy. The presence of the disorder can then be confirmed by genetic testing. The genetic test requires a sample of blood from the patient. The DNA is then extracted from the blood and analyzed to see if that person has the mutation that causes myotonic dystrophy. Prenatal testing, where the DNA of the fetus is checked for the presence of the myotonic dystrophy mutation, is also available.

Diagnosis of myotonic dystrophy is not difficult once the disorder is suspected. However, delays in diagnosis are common. More common diseases with symptoms that mimic myotonic dystrophy must typically first be ruled out before this disorder is considered. The symptoms are complex. Physicians may see only one or two patients with DM in their entire practice and may not be familiar with the range of ways this disease can present.

What is the prognosis for myotonic dystrophy diagnoses?

Myotonic dystrophy is a progressive or degenerative disease. Symptoms tend to worsen gradually over several decades. While no treatment exists that slows the progression of myotonic dystrophy, management of its symptoms can greatly improve patient quality of life. Early intervention can reduce or avert complications that sometimes arise.

DM2 tends to be less severe than DM1 and has minimal impact on life expectancy. DM1 is much more variable and the prognosis for an affected individual is difficult to predict. Some people may experience only mild stiffness or cataracts in later life. In the most severe cases, respiratory and cardiac complications can be life-threatening even at an early age. In general, the younger an individual is when symptoms first appear, the more severe symptoms are likely to be.

However, prognosis is as variable as the symptoms of this disease. How myotonic dystrophy affects one individual can be completely different from how it manifests in another, even for members of the same family. It is impossible to predict how the disease will affect any single individual.

What DM treatment or therapies are available?

No treatments currently exist that slow the progression of myotonic dystrophy, but symptomatic treatments are available. Managing the symptoms of this disease can reduce suffering and improve quality of life for patients. Ongoing monitoring can avert or reduce the complications seen at critical times.

Note: Medical information available on this site is designed as general information only. Patients should consult a physician or other qualified medical professional for advice on medical treatment.

Symptomatic treatments:

Medications

- Anti-diabetic drugs to normalize blood sugar levels and address mild diabetic symptoms
- Anti-myotonic drugs (such as mexiletine) when myotonia impairs normal activities
- Nonsteroidal anti-inflammatory drugs to manage muscle pain

Rehabilitative therapy

- Physiotherapy for muscle weakness, myotonia and contractors
- Speech therapy for swallowing and pronunciation issues
- Psychiatric therapy for behavioral and psychological issues (such as attention deficit, depression and anxiety disorders)
- Individualized support for learning disabilities and cognitive delays

Devices

- Assistive devices (such as neck braces, arm and foot braces, canes, walkers, scooters, and wheelchairs) to ensure safe navigation
- Eye crutches for droopy eyelids (ptosis)
- Pacemaker or implantable cardioverter defibrillator (ICD) to address irregular heartbeat issues
- Incentive spirometry and cough assist devices to improve respiratory function
- Continuous positive airway pressure (CPAP) device to ensure respiratory sufficiency

Surgery

- Orthopedic surgery for gait issues and contractors
- Cataract removal
- Eyelid surgery to correct droopy eyelids

However, surgery is typically used as a last resort treatment as DM patients have an elevated risk of complications associated with the use of anesthesia.
Please review the Anesthesia Guidelines for further information.

If two siblings have the disease, will they have similar organ issues over time?

Not always. Their genetic background is different although many genes are shared. The genomic background is likely to play an important role in organ-specific phenotype expression.

If you know you have the disease, are there ways to have children who are not affected? Are there any additional risks for an affected woman during pregnancy? Are there any precautions she should take if she becomes pregnant?

PGD and prenatal screening will be useful, if the couple accepts the ethical implications. Risks for the pregnant woman consist of polyhydramnios, placenta previa, miscarriage, preterm birth, stillbirth, and complications in labor and delivery including prolonged labor, anesthesia risks and postpartum hemorrhage. Psychological risks, especially after having a baby with the congenital form, should be considered.

Is there always an expansion at every generation? Is it larger with maternal transmissions? Or is there an identical distribution between men and women?

Not always. About 6% of paternal transmissions result in contraction of the repeat in the offspring. Expansion is more prominent with paternal transmissions when the repeat is small (37-100) while it is much larger with maternal transmissions when the repeat is over a few hundred.

Is there new information on DM impact on the brain and how executive function is affected, especially in childhood onset?

There in only one paper that introduced a new concept that assesses the progressive nature of cognitive dysfunction in some childhood-onset patients (Echenne, et al. 2007 Eur J Paediatr Neurol). Several research centers, including the University of Minnesota, are keenly interested in brain involvement as an aspect of DM.

Regarding anesthetic risks, what specifically should DM patients tell an anesthesiologist before surgery?

Perioperative complication is increased in patients with DM. All drugs, including sedatives, induction drugs, anesthetics, neuromuscular junction blockers and opiates must be carefully chosen, and doses must be carefully determined. In particular, anticholiesterases (e.g. neostigmine), depolarizing neuromuscular blocking agents (e.g., suxamethonium) and inhalational anesthetics should be avoided. Cardiac problems should be alerted to the anesthesiologist, who should also be aware that hyperkalemia, hyperthermia and shivering and mechanical or electrical muscle stimulation can cause myotonia, which may interfere with the surgery. Perioperative aspiration is a risk due to bulbar weakness.

If one develops serious cardiac problems - a very rapid or very slow heartbeat, or arrhythmia (irregular heartbeat) - what short of device is generally implanted in one's chest? How should a DM patient be followed from a cardiac standpoint (e.g. EKGs, echos, etc.)?

The patient should have EKG, echo and electrophysiological (EP) studies, depending on the nature of arrhythmia to determine the need for a pacemaker. If a pacemaker needs to be implanted, a device with pacemaker/defibrillator capability may be preferable.

Many patients have many problems with diarrhea and constipation. Are these related to the following: a) digestion, b) type of food eaten, c) muscles not working properly? How can these problems be treated?

Most problems are due to intestinal motility. Selection of foods is important. Appropriate amounts of fiber supplements may be useful, although overuse may produce impaction if the patient becomes constipated. Stool softener and non-irritant laxatives are useful. Cisapride (Propulsid) and other prokinetic drugs should be avoided because of cardiotoxicity.

What does one do when swallowing becomes a problem? What emergency interventions should be followed? Does chewing food a lot help food go down easily? Does drinking lots of liquids with a meal help? Any particular type of liquid?

Swallowing problems come from both oropharyngeal muscle weakness and abnormal motility of the esophagus. The patient should be evaluated by a speech pathologist and a gastroenterologist with fibroscopic and manometric testing. If patients are not able to do these studies, the modified barium swallow test should be done to assess the risk for aspiration. The speech therapist should be able to give useful advice to alleviate the problem. If the problem imposes high aspiration risks, G tube insertion should be considered. Chewing, drinking fluids, and pureed foods may help. If aspiration occurs, or may have occurred and fever starts, go to an emergency center for treatment.

What can be done to help patients who need ventilators to establish a beneficial level of support?

For those who have a tracheotomy, the right answer may be "as needed" if the patient can get off the ventilator intermittently. Keeping the patient comfortable and able to communicate is important.

Recognizing that exercise does not prevent the progression of muscle weakness in DM, are there exercise regimens that are recommended to try and maintain what muscle strength is present?

Overdoing is counter-productive. Low intensity aerobic training may be useful.

How reliable is pre-implantation DNA analysis of embryos and is there information on rate of successful pregnancy when the woman does not have DM?

It is increasingly available. Kakourou et al. "Preimplantation genetic diagnosis for myotonic dystrophy type 1 in the UK," *Neuromuscular Disorders* is the newest paper available.

Is there a medication to help with daytime sleepiness? What is the relationship between sleep apnea and DM?

Modafinil is the choice although it is expensive. Sleep apnea contributes to the daytime somnolence but patients often continue to have daytime sleepiness after CPAP.