A Guide for Patients and Families

What is...

Myotonic Dystrophy

Myotonic dystrophy (DM) is a **musculoskeletal disorder** that affects the muscles and a number of different organs in the body.

DM is the most common form of muscular dystrophy that **begins in adulthood,** usually in the 20s or 30s.

It is a **genetic disorder** characterized by progressive muscle loss and weakness.

DM causes weakness of the **voluntary muscles,** although the degree of weakness and the muscles most affected vary greatly according to the type of DM and the age of the person with the disorder.

DM is classified into two types, **type 1** and **type 2,** each of which may affect different muscles.

People with DM often experience prolonged **muscle tensing (myotonia)** and are not able to relax certain muscles after use. For example, someone with DM may have difficulty letting go of someone's hand after shaking it.

The severity of the disease may vary, even among members of the same family. However, in general, symptoms tend to progress slowly.

Type 1 DM (DM1) occurs when a gene on chromosome 19 called **DMPK** contains an abnormally expanded section.

Type 2 DM (DM2) is caused by an abnormally expanded section in a gene on chromosome 3 called **ZNF9.**

In some cases, babies are born with a variation of myotonic dystrophy type 1 called congenital myotonic dystrophy.

DM is inherited in an **autosomal dominant** manner, which means it takes the mutated (flawed) gene from only one parent to cause the disease.

There is no cure for DM, but medications and therapy can help manage some of its symptoms.

MDA® Muscular Dystrophy Association

What are the signs and symptoms of DM?

DM is a multi-system condition affecting many parts of the body. The symptoms in people with DM2 tend to be milder than in those with DM1, but the symptoms may overlap.

People with DM1 typically experience involvement of the legs, hands, neck, and face, while individuals with DM2 typically experience involvement of the neck, shoulders, elbows, and hips.

Nervous system
- Cerebral atrophy
- Developmental delay
- Excessive daytime sleepiness
- Facial diplegia

Cognition
- Intellectual disability
- Learning disability
- Obsessive-compulsive traits

Gastrointestinal
- Dysphagia
- Feeding difficulties in infancy

Hormonal/sexual
- Complicated pregnancy
- Male baldness
- Male infertility
- Low sperm count
- Testicular degeneration

Skeleton and muscle
- Hypotonia
- Muscle weakness
- Muscle stiffness
- Muscle tightness
- Muscle wasting
- Myalgia
- Myotonia

Heart
- Cardiac conduction defects
- Cardiomyopathy
- Palpitations
- Tachycardia

Respiratory system
- Breathing difficulties

Endocrine
- Diabetes

What should I know about DM?

1. Overall intelligence is usually normal in people with DM, but learning disabilities and an apathetic demeanor are common in the type 1 form.

2. Generally, the earlier DM1 begins, the more profound the symptoms tend to be. In congenital DM1, which affects children from the time of birth, there can be serious impairment of cognitive functioning. These children also may have problems with speech, hearing and vision.

3. Children with congenital-onset DM1, once they survive the crucial neonatal period of respiratory muscle weakness with the help of assisted ventilation, usually show improvements in motor and breathing functions. They may have cognitive impairment, delayed speech, difficulty eating and drinking, and various other developmental delays.

4. The childhood-onset form of DM1 — beginning after infancy but before adolescence — is more often characterized by cognitive and behavioral abnormalities than by physical disabilities. Eventually, muscle symptoms develop, to varying degrees.

5. The most common type of DM1 — the adult-onset form — begins in adolescence or young adulthood, often with weakness in the muscles of the face, neck, fingers, and ankles. The weakness is slowly progressive for these and eventually other muscles.

6. As the disease progresses, the heart can develop an abnormal rhythm and the heart muscle can weaken. The muscles used for breathing can weaken, causing inadequate breathing, particularly during sleep.

7. In DM1, the involuntary muscles, such as those of the gastrointestinal tract, can be affected. Difficulty swallowing, constipation, and gallstones can occur. In females, the muscles of the uterus can behave abnormally, leading to complications in pregnancy and labor.

8. Life expectancy may be reduced for people with DM1. An increased risk of death may be associated with younger age of onset, more severe muscle weakness, and cardiac conduction defects. People with more mild symptoms of DM1 may have a normal lifespan.

9. DM2 is, in general, a milder disease than type 1. It does not appear to have a congenital-onset form and rarely begins in childhood.

10. In contrast to type 1 DM, the muscles affected first in DM2 are the proximal muscles — those close to the center of the body — particularly those around the hips. However, some finger weakness may be seen early as well. The disorder progresses slowly, but mobility may be impaired early because of weakness of the large, weight-bearing muscles.

11. Definitive information about prognosis in people with myotonic dystrophy type 2 is limited, but the condition generally runs a milder course. People with DM2 may have a normal lifespan. While mobility may be impaired at an early age, the ability to walk is often retained until around 60 years old.

How is DM treated?

There currently is no cure or specific treatment for DM. Treatment is based on each person's specific signs and symptoms.

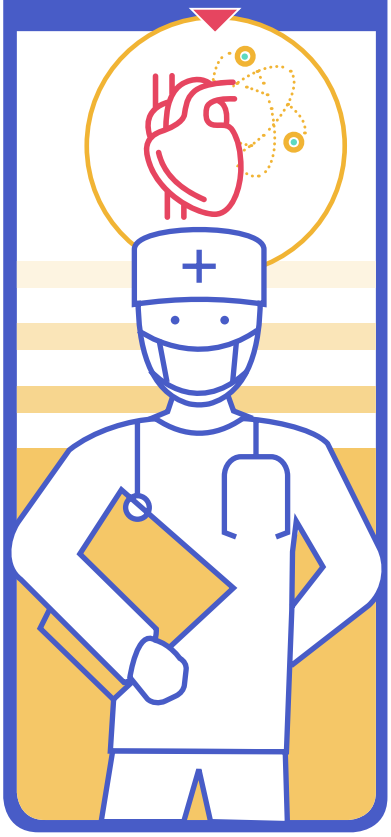
Physical therapy helps to restore and maintain muscle strength and function through exercise, as well as to maintain range of motion through stretching.

Routine physical activity may help control musculoskeletal pain and maintain muscle strength and endurance.

Educational and psychological interventions can help with learning disabilities.

Supportive aids such as braces, walkers, or wheelchairs may help those who experience problems with muscle weakness or fatigue.

Pacemakers and implantable defibrillators can be used to treat abnor



Occupational therapy can help improve daily living and work skills.

Surgery can be used to remove cataracts.

Assisted ventilation can help treat respiratory muscle weakness.

Cough assistance machines and assisted cough techniques can help people clear out secretions from the lungs.

Treatment with **angiotensin converting enzyme (ACE) inhibitors** and beta blockers may be used to help lessen stress on the heart.

Heat and **massage** may help relieve muscle pain.

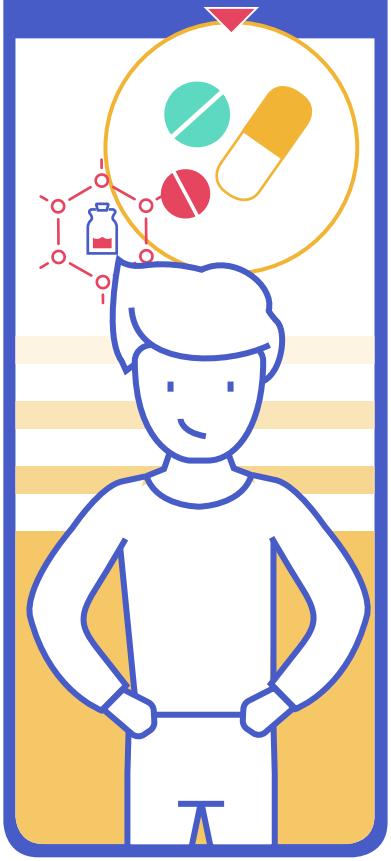
Medications that increase alertness may be used to treat excessive daytime sleepiness.

Laxatives, suppositories, or **enemas** may be used to help manage constipation.

Gallbladder removal may relieve persistent abdominal pain caused by gallstones.

Testosterone replacement therapy may be used to treat infertility in males.

Over-the-counter pain remedies or prescription pain medications may be used to reduce pain in the skeletal muscles.



Please talk to your medical provider to obtain more information on these treatments.

MDA Glossary

Atrophy
A decrease in the size and mass of muscle tissue

Cardiomyopathy
A condition in which the heart muscle is weakened, making it harder for the heart to pump blood to the body

Cerebral atrophy
Degeneration of the cerebrum

Conduction defect
Irregular electrical control of the heartbeat

Dysphagia
Difficulty swallowing

Facial diplegia
Paralysis of muscles in the face

Hypotonia
Decreased muscle tone

Muscular dystrophy
A term that refers to a number of diseases that cause progressive loss of muscle mass, resulting in weakness and, sometimes, loss of mobility

Mutation
A flaw in the DNA code

Myalgia
Muscle pain

Myotonia
The inability to relax muscles at will

Palpitation
Missed heartbeat

Tachycardia
Fast heartbeat

To learn more about DM, visit mda.org or contact the MDA National Resource Center at 833-ASK-MDA1 (275-6321).

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