

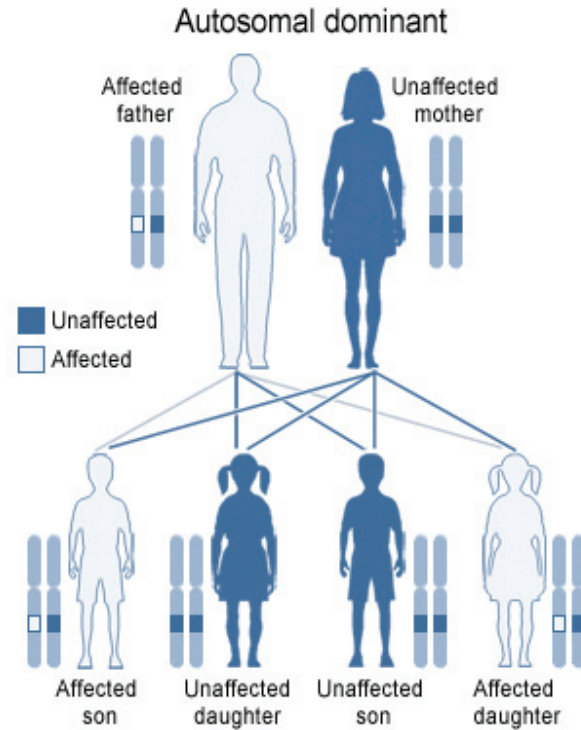
Genetics and Testing

Myotonic Dystrophy is caused by an expansion of part of the chromosomes, either Chromosome 19 for DM1 and CMYD, or Chromosome 3 for DM2. There is an excess of triple repeat CTG on the chromosome in DM1 or CCTG in DM2. Normally, a person has 1–49 repeats, but a person with DM has 50 to 2,500 or more repeats. Myotonic Dystrophy is autosomal dominant meaning the disease runs in families and each child has a 50% chance of having the disease if one parent has the disease. Myotonic dystrophy has an aspect called anticipation, which means that each succeeding generation has the disease slightly more severe. Recent research indicates that the excess number of repeats on the chromosome interferes with the normal process of the body synthesizing mRNA, which is the template for the bodies proteins. The DM1, DM2 and CMYD can be diagnosed with a DNA test. Testing is important for individuals that may have any type of myotonic dystrophy. Many individuals with DM will have a reaction to general anesthesia, even those that have no other visible symptoms, so testing is important. Women in childbearing age may transmit the Congenital form of the disease, although they may not have any visible form of the disease.

Testing is available from several resources, including Athena Diagnostics. 1-800-394-4493

Additional Genetic Resources

<http://neuromuscular.wustl.edu/>



Myotonic Dystrophy is so variable that some people may not know they have it, while others are severely affected. The disease is slowly progressive, but many of the symptoms can be managed.

There are drugs that can help with muscle pain and sleep disorders. We urge you or your caregiver to actively seek aggressive testing and treatment. Your local Muscular Dystrophy Association clinics have a team of specialists to help you with the disease.

Visit our website for more information. Carrying our plastic medical alert card is important for all Myotonic Dystrophy patients. Call us, write to us or email your request with your full address.

Email: info@myotonicdystrophy.org



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Myotonic Dystrophy

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Leading the way to management and treatment

General Information on DM1 and DM2

This pamphlet is written for individuals and families with Myotonic Dystrophy, also known as Steinert's Disease or Dystrophia Myotonica (DM). It describes the types of DM, what causes DM, and some management techniques.



DM is the most common inherited muscle disease, affecting about one in every 8,000 people. In general, DM consists of muscle weakness and myotonia (slow relaxation of muscles after contraction), which gets more severe over time. Specific problems in other systems of the body can also occur.

Since DM can affect many tissues and organs, it is called a "multisystemic" disorder.

Myotonic Dystrophy is an extremely variable condition. It can vary in severity, in the systems of the body it affects and in the age of onset, even in the same family. People with milder symptoms may never be diagnosed as having DM.

Helpful Resources

www.myotonicdystrophy.org
www.mdausa.org
www.athendiagnosics.com

On the other hand, newborn babies who are more severely affected may die during infancy without the diagnosis of DM ever having been made. If a newborn is diagnosed with DM, this may be the first time the family learns about the disease. The discovery of the gene alteration, which causes DM, helps explain this condition's incredible variation.

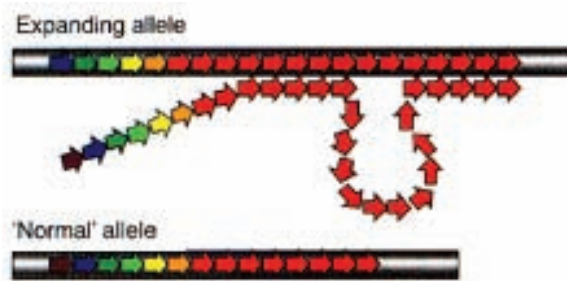
Types of Myotonic Dystrophy

There are several types and subtypes of Myotonic Dystrophy. Although there is no cure for DM, its symptoms can be managed.

Myotonic Dystrophy or DM1

The first identified type of Myotonic Dystrophy is DM1, which has an incidence of 1 in 8,000. This disease can occur at several stages: juvenile onset, early adult onset and late adult onset. The disease may cause all or just a few of these symptoms:

- Slow relaxation of muscles after contraction
- Muscle stiffness
- Facial muscle weakness
- Eye muscle weakness
- Jaw muscle weakness
- Weakness of distal muscles: forearm, hands, feet and ankles.



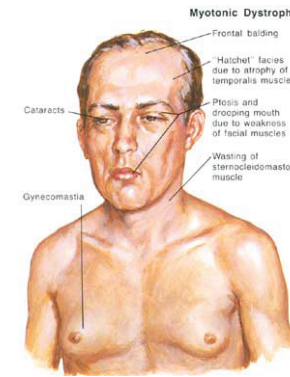
- Speech problems
- Swallowing difficulty
- Irregular heart beat
- Diabetes
- Testicular atrophy
- Hair loss (men & women)
- Cataracts
- Problems with excessive sleepiness
- Apathetic attitude, lack of energy

The surprising aspect of DM1 is that the symptoms can be variable. One person may have the disease in a very mild form. Others may be severely affected. The disease is slowly progressive so over the years the symptoms will worsen. The disease also tends to get worse with each succeeding generation. So the children which are affected will have the disease more severely than the parents.

Precautions must be taken for anyone with DM1 that will have anesthesia. It is particularly important to have periodic health check-ups. Most of the symptoms can be managed by properly trained medical staff.

Myotonic Dystrophy Type 2 or DM2

DM2 has many of the symptoms of DM1, but the genetic problem is on Chromosome 3. DM2 is also known as PROMM (Proximal Myotonic Myopathy). People who have DM2 may not have problems with sleepiness associated with DM1. The muscle weakness is in the trunk (proximal muscles). The incidence of DM2 has been estimated at 1 in 10,000 and there are diagnostic tests available. A support group and extensive information is available through PROMM at Yahoo Groups.



Congenital Myotonic Dystrophy or CMyD

Congenital DM refers to the presence of symptoms in the first month of life. The symptoms may include facial weakness, hypotonia (low muscle tone), respiratory problems, feeding difficulties and talipes (clubfoot). Delays in motor skills such as rolling over, crawling, and walking usually occur. Some degree of mental impairment may occur in individuals with CMyD.

For reasons not yet clearly understood, Congenital DM almost never occurs unless the baby's mother has DM herself. Children may make considerable progress from birth to age 4-5 or so. However, children with this condition acquire the adult form in their teens.

Treatments

There is no cure yet for Myotonic Dystrophy, but several promising approaches are under investigation. For example, Iplex from Insmed is in Phase 2 trials now.

Until specific therapies are cleared for treatment the recommendations are to treat the individual symptoms as they arise.

Aggressive treatment for heart relates issues, pneumonia or lung infections is important. In the USA the Muscular Dystrophy Association is very helpful with local and regional clinics.