

Myotonic Dystrophy (Steinert's disease)

What is myotonic dystrophy?

Myotonic dystrophy, also known as Steinert's disease, is the most common form of muscle disease, affecting approximately one person in 8,000 worldwide. It is a disorder characterized by progressive muscle weakness and wasting and by myotonia (difficulty in relaxing the muscles after they have been contracted). It is a multi-system disease, typically involving a wide range of other tissues as well as muscle.

Common symptoms in the adult form include:

- myotonia that results in a delay in the ability to relax the muscles after a prolonged contraction,
- muscle weakness of the voluntary muscles, starting gradually and progressing slowly,
- muscle stiffness,
- drooping eyelids,
- unclear pronunciation of words,
- difficulty raising the head when lying,
- difficulty holding an object firmly or lifting it,
- a shuffling gait when walking,
- difficulty climbing stairs or getting up from a seated position
- a long, rather expressionless face.

People with myotonic dystrophy may also have symptoms affecting other parts of the body.

Who can be affected by myotonic dystrophy?

Anyone can be affected by myotonic dystrophy. It is a genetic disorder passed on to children of either sex by one parent who has the disorder.

Myotonic dystrophy can affect people at any age. The majority of people are diagnosed by the time they reach their early twenties. With each successive generation, the symptoms of myotonic dystrophy seem to get more severe, and the age that they appear gets younger. This phenomenon is known as anticipation.

What are the different forms of myotonic dystrophy?

The adult form of myotonic dystrophy generally appears between the ages of 10 and 30 years. The severity of symptoms, the rate of progression and the degree of disability vary widely from one person

to the next, even among members of the same family. In most cases, the disorder progresses slowly.

Congenital myotonic dystrophy, the most severe form of myotonic dystrophy, is always present at birth. Affected babies are born to mothers who also have myotonic dystrophy. When the father has myotonic dystrophy, his children are not at risk for the more serious congenital form. Babies with congenital myotonic dystrophy are very weak and floppy and frequently have problems with sucking, feeding and breathing. If they survive the newborn stage, they generally overcome their breathing and

feeding problems, but they are slow to develop language and motor skills and are often affected for life with difficulties in these areas.

What causes myotonic dystrophy?

Myotonic dystrophy is caused by an error in genes located on chromosome 19 or chromosome 3. The basic fault in DM1 is an unstable inherited mutation in the myotonic dystrophy protein kinase gene. At the present time, it is not known how the genetic abnormality causes the disorder.

How is myotonic dystrophy inherited?

Myotonic dystrophy is transmitted via an autosomal dominant pattern of inheritance. Autosomal refers to the fact that the faulty gene appears on one of the 22 chromosomes not associated with determining the sex of the child (in this case, chromosome 19). Dominant refers to the fact that the disorder is passed down by one parent of either sex who also has the disorder. There is no carrier status. Each child born to an affected parent has a 50% chance of inheriting the disorder.

How is myotonic dystrophy diagnosed?

A physician makes a diagnosis based on family history and a physical examination. Tests that will assist the physician in making his diagnosis include DNA analysis an electromyogram (to measure electrical activity in the muscle), a muscle biopsy (to study muscle cells for signs of the disease) and in the case of possible cataracts, an eye examination.

Is there any cure or treatment for myotonic dystrophy?

There is no cure for myotonic dystrophy at the present time. Treatment is symptomatic. That is, problems associated with myotonic dystrophy are treated individually. For example, surgery is available for correction of cataracts. Medication may be prescribed to counter the effects of myotonia. A heart specialist, depending on what symptoms are experienced, will treat heart problems. Speech therapy and a modified school environment can assist children with developmental delays. Remaining as active as possible is recommended for everyone with myotonic dystrophy.

What are the symptoms of myotonic dystrophy?

Cataracts may develop frequently in people with myotonic dystrophy. They develop fairly slowly, but can occur in people as young as 30 years.

Myotonic dystrophy may affect the heart muscle. A person may experience palpitations (rapid, bounding pulse) or dizzy spells, or they may have no symptoms whatsoever.

A person who has myotonic dystrophy may have difficulty swallowing. This is due to involvement of smooth (or involuntary) muscle. Cold foods may cause some individuals to choke.

Other potential problems may include bowel problems (constipation and stomach pain) and uterine problems in females. Affected individuals may be susceptible to respiratory problems such as infections and shortness of breath.

Premature balding may occur in some males, while females may experience thinning of their hair. In addition to the symptoms of the adult form, symptoms of the congenital form of myotonic dystrophy include: difficulty breathing, sucking and/or feeding, weakness in virtually all muscles and slowness and difficulty in developing language and motor skills.

What about research?

Researchers are trying to determine how the expansion of the repeated section of DNA in genes on chromosome 19 or chromosome 3 destabilizes muscle cell function. Other scientists are studying the actual process of muscle degeneration in myotonic dystrophy and also, the effect of myotonic dystrophy on other parts of the body.

How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

**All Muscular Dystrophy Canada Information
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