

Myotonic Dystrophy

➤ Myotonic dystrophy (DM) is a complex multisystem genetic disorder that may affect multiple organs, including; skeletal muscle, the eye, heart, brain, hormone levels, the gastrointestinal tract, the uterus and skin.

➤ Myotonic dystrophy is a genetic disorder. Two major types are recognized (DM1 and DM2). DM1 & DM2 have different genetic causes and a somewhat different pattern of symptoms and characteristics. DM2 appears to be less common and also milder than DM1.

➤ Congenital Myotonic Dystrophy (CMD): symptoms appear in the newborn, this is a rarer but far more severe form of the same mutation that causes DM1 (it is not seen in DM2).

➤ Myotonic dystrophy is the second most common muscular dystrophy (MD) overall (behind Duchenne MD) but the most common adult form of muscular dystrophy. Affects about 1 in 8,000 people worldwide but it also occurs in “pockets.” DM1 is disproportionately common in the Saguenay-Lac-Saint-Jean region of Quebec with about 1 in 500 people affected.

➤ Symptoms: Myotonia involves abnormally long muscle contractions or a slowed relaxation after a muscle contraction as shown by an inability to relax voluntary muscles at will. The core features of myotonic dystrophy are myotonia of the muscles and weakness of the distal muscles – starting in the wrists and fingers, neck and face (eye musculature, tongue and lips) and the lower legs and ankles. Later, weakness spreads to the thighs, hips and shoulders. Chest muscles and the diaphragm may also become involved. Cataracts are common and a wide range of other symptoms may also appear. Myotonic dystrophy is an extremely variable condition and symptoms can vary widely in severity, even within the same family. About 50% of people with the disorder show some signs by age 20, but many do not develop clear symptoms until after 50.

➤ The more severe the mutation involved, the more severe the impact will be (especially true of DM1). Myotonic dystrophy generally becomes progressively more disabling with time and the impact on other organs and body systems may also increase over time.

➤ There is evidence that myotonic dystrophy affects the brain and often has a deleterious effect on intelligence as measured by IQ and on memory tasks. Also, certain personality traits have been associated with myotonic dystrophy, including a lack of motivation, lack of insight and poor judgment, and avoidant, obsessive-compulsive and passive-aggressive traits.

➤ Cause: Muscle weakness and other problems are caused by known genetic mutations; however, the exact mechanisms involved are not well understood. DM1 is caused by a mutation that expands a section of DNA code (a CTG repeat) in the DMPK gene located on Chromosome #19. People normally have about 5 to 30 CTG repeats in this section. Mild DM1 is seen when there are from 50 to 80 repeats present and severe cases may show more than 2000 repeats. Congenital DM1 may involve more than 4000 repeats. DM2 is caused by a CCTG expansion mutation in the ZNF9 gene, found on chromosome #3, involving an expansion with from 75 to 11,000 repeats and a mean of about 5,000 repeats.

➤Classification of DM based upon severity:

- 1). “Mild DM” (adult onset): People with “mild DM” often lead active lives and may even be unaware that they have the disorder, attributing their stiffness to “arthritis.”
- 2). “Classical DM” (adult onset): People commonly have muscle weakness and wasting, myotonia, hand and wrist weakness and/or foot drop.
- 3). “Congenital Myotonic Dystrophy” (CMD): A very severe form of DM1, often fatal in young children, surviving children will display classical DM1 symptoms as they grow up.

➤Onset: Typically, in the most common form, symptoms appear in the second or third decade of life, although a mild mask-like appearance of the face (“myotonic facies”) and some myotonia may often be observed in childhood.

➤Diagnosis: Some symptoms of myotonic dystrophy might be hard to see in mild cases. In typical adult-onset cases, clinical diagnosis is usually straightforward, presenting with progressive distal weakness in the presence of myotonia, with frontal balding, and cataracts.

➤Genetic Testing: DNA tests can show the presence and size of the mutation (the expansion of a repeat) and permit DNA diagnosis of DM (including prenatal diagnosis).

➤Myotonic dystrophy is inherited in an autosomal dominant fashion although some rare cases may spontaneously appear due to new mutations occurring in the early stages of embryonic development.

➤There are currently no treatments for myotonic dystrophy that can halt or reverse the symptoms and muscle weakness. Each symptom displayed reflects an underlying issue and each needs to have a treatment plan developed to best address the concern on an individual basis.

➤ANESTHESIA WARNING: An unusually high rate of complications and even deaths are associated with general anesthesia in patients with DM. Even mild myotonic dystrophy can cause very serious complications. Mild cases may also be even more at risk because patients may not think to tell their surgeons about their diagnosis or a family history of myotonic dystrophy.

Neuromuscular disorders and what people commonly call muscular dystrophies are very complex and often devastating conditions. Some strike children, others do not display symptoms until well into adult life. Most are progressively disabling, getting worse over time and some are fatal, often in childhood. Some affect primarily muscles, others can affect a wide spectrum of health, including; personality and intellectual functioning, the eyes, the bones, digestion and diabetes to list just a few. There are over 100 different types of disorders and variations listed under this umbrella. Many disorders have genetic causes but in many cases, just how these genetic mutations cause the symptoms we see is not yet understood. Every day, medicine is making progress in understanding these disorders, but at the present time, there are more theories than facts. Sadly there are no direct treatments yet.

Please feel free to contact Muscular Dystrophy Canada if you have suggestions or questions about this page. Our website is: <http://www.muscle.ca/> or e-mail us at info@muscle.ca

Thank you.