



## Myotonic Dystrophy type 2

Other Names: Proximal Myotonic Myopathy

*Myotonic Dystrophy type 2 is a genetic condition that affects the muscles and several other body systems. It is caused by mutations in the CNBP gene which makes a nucleic acid binding protein.*

### Characteristics of Myotonic Dystrophy type 2

It is estimated that about 1 in 8000 individuals are affected with myotonic dystrophy. There are two types of myotonic dystrophy called myotonic dystrophy type 1 (DM1) and myotonic dystrophy type 2 (DM2). These two types share similar symptoms, but they have a different genetic cause.

DM2 is a multisystem disease, meaning that it affects several organs in the body. Specifically, myotonic dystrophy type 2 is associated with muscle problems (i.e., muscle pain and myotonia (muscle stiffness or the inability of muscles to relax)), heart problems (i.e., cardiac arrhythmias (irregular heart rhythms) or cardiomyopathy (weakening of the heart)), cataracts, and endocrine problems (e.g., diabetes). Males with DM2 also have infertility. In general, most individuals with DM2 shows signs or symptoms of the disease in their 30s or 40s, however there are reports of symptoms being present from childhood to late adulthood. The age symptoms present and what symptoms affect an individual is different among family members.

### Diagnosis/Testing

Individuals with DM2 have a change or mutation in a gene called CNBP. This gene contains a four-letter code, CCTG, that is repeated over and over again, and thus it is known as a "CCTG repeat." The number of CCTG repeats can be different from one person to another. Individuals who do not have DM2 usually have less than 44 CCTG repeats. However, individuals with DM2 usually have an abnormally high number of CCTG repeats that can range from over 75 to several thousand repeats. A large number of CCTG repeats is known as an expanded CCTG repeat allele. The CCTG repeat expansion is thought to disrupt a cell function called splicing (i.e., a process by which the cell makes different versions of a protein). Each version of the protein has a separate function. If splicing is disrupted, the cell will have too much of one version of the protein and not enough of another. This leads either to cell dysfunction or cell death. The CCTG repeat is very unstable and has been seen to get larger as person ages. It has also been seen to change in size when passed from generation to generation. However, the size of the CCTG expansion cannot predict the age of symptoms or what type of symptoms will present in an affected individual.

### Management/Surveillance

Management of DM2 often involves physical and occupational therapy to manage pain and adapt to muscle weakness; treatment of diabetes; removal of cataracts; and testosterone replacement therapy for males with DM2. Individuals with DM2 often require tests (e.g., EKG and echocardiogram) to detect and monitor for heart problems, and blood tests to screen for diabetes.

Females with DM2 who are pregnant may have an increase in their muscle symptoms including myotonia, fatigue and muscle weakness. These symptoms tend to lessen after the pregnancy.

## Mode of inheritance

Myotonic dystrophy type 2 is inherited in an autosomal dominant pattern. This means inheriting one CNBP CCTG expanded repeat allele is enough for an individual to be affected and show signs of DM2. The mutation is inherited from an affected parent and affects males and females equally.

## Risk to family members

If a parent has a CNBP CCTG expansion, the risk of having a child with DM2 is 50% with each pregnancy. The risk to develop symptoms and the age of onset cannot be predicted by CCTG repeat size or the clinical symptoms of other family members.

## Special considerations

None

## Resources

Muscular Dystrophy Association (MDA)

<http://www.mdausa.org/>

Myotonic Dystrophy Foundation

<http://www.myotonia.org>

Genetics Home Reference: Myotonic dystrophy

<http://ghr.nlm.nih.gov/condition/myotonic-dystrophy>

## References

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**Created by:** Joline Dalton, MS, CGC

**Updated:** mm/yyyy

**Edited by:** Seema Jamal, MSc, LCGC