DMPK gene

DM1 protein kinase

Normal Function

The *DMPK* gene provides instructions for making a protein called myotonic dystrophy protein kinase. Although the specific function of this protein is unknown, it appears to play an important role in muscle, heart, and brain cells. This protein may be involved in communication within cells. It also appears to regulate the production and function of important structures inside muscle cells by interacting with other proteins. For example, myotonic dystrophy protein kinase has been shown to turn off (inhibit) part of a muscle protein called myosin phosphatase. Myosin phosphatase is an enzyme that plays a role in muscle tensing (contraction) and relaxation.

One region of the *DMPK* gene contains a segment of three DNA building blocks (nucleotides) that is repeated multiple times. This sequence, which is written as CTG, is called a triplet or trinucleotide repeat. In most people, the number of CTG repeats in this gene ranges from 5 to 34.

Health Conditions Related to Genetic Changes

Myotonic dystrophy

Type 1 myotonic dystrophy results from a mutation in the *DMPK* gene known as a trinucleotide repeat expansion. This mutation increases the size of the repeated CTG segment in the *DMPK* gene. People with type 1 myotonic dystrophy have from 50 to 5,000 CTG repeats in most cells. The number of repeats may be even greater in certain types of cells, such as muscle cells.

The mutated *DMPK* gene produces an altered version of messenger RNA, which is a molecular blueprint of the gene that is normally used to guide the production of proteins. Researchers have found that the altered messenger RNA traps proteins to form clumps within the cell. The clumps interfere with the production of many other proteins. These changes prevent muscle cells and cells in other tissues from functioning properly, leading to muscle weakness and the other features of type 1 myotonic dystrophy.

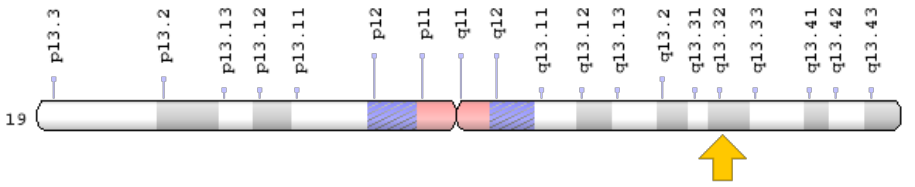
The size of the trinucleotide repeat expansion is associated with the severity of signs and symptoms. People with the classic features of type 1 myotonic dystrophy, including muscle weakness and wasting beginning in adulthood, usually have between 100 and 1,000 CTG repeats. People born with the more severe congenital form of type 1 myotonic dystrophy tend to have a larger number of CTG repeats, often more than 2,000. This form of the condition is apparent in infancy and may involve life-threatening health problems.

As the altered *DMPK* gene is passed from one generation to the next, the size of the CTG repeat expansion often increases in size. People with 35 to 49 CTG repeats have not been reported to develop type 1 myotonic dystrophy, but their children are at risk of having the disorder if the number of CTG repeats increases. Repeat lengths from 35 to 49 are called premutations.

Chromosomal Location

Cytogenetic Location: 19q13.32, which is the long (q) arm of chromosome 19 at position 13.32

Molecular Location: base pairs 45,769,709 to 45,782,557 on chromosome 19 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DM-kinase
- DM-PK
- DM protein kinase
- DM1
- DM1PK
- Dm15
- DMK
- DMPK_HUMAN
- dystrophia myotonica 1
- dystrophia myotonica kinase, B15
- dystrophia myotonica protein kinase
- dystrophia myotonica-protein kinase
- MDPK
- MT-PK

- myotonic dystrophy protein kinase
- myotonin
- myotonin-protein kinase

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: DM1 Mutation Is an Expansion of CTG Trinucleotide Repeats
  https://www.ncbi.nlm.nih.gov/books/NBK6512/#A5300

Clinical Information from GeneReviews

- Myotonic Dystrophy Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1165

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DMPK%5BTIAB%5D%29+OR+%28dystrophia+myotonica-protein+kinase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- DYSTROPHIA MYOTONICA PROTEIN KINASE
  http://omim.org/entry/605377

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DMPK.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DMPK%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2933
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1760
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1760
- UniProt
  https://www.uniprot.org/uniprot/Q09013

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