DMPK gene

DM1 protein kinase

Normal Function

The *DMPK* gene provides instructions for making a protein called myotonic dystrophy protein kinase. This protein appears to play an important role in muscle, heart, and brain cells. The 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

Mutations in the *DMPK* gene cause a form of myotonic dystrophy known as myotonic dystrophy type 1. Myotonic dystrophy is characterized by progressive muscle wasting and weakness. The muscle weakness associated with type 1 particularly affects muscles farthest from the center of the body (distal muscles), such as those of the lower legs, hands, neck, and face. People with this disorder often have prolonged muscle contractions (myotonia) and are not able to relax certain muscles after use.

The type of mutation that causes myotonic dystrophy type 1 is known as a trinucleotide repeat expansion. This mutation increases the size of the repeated CTG segment in the *DMPK* gene. People with myotonic dystrophy type 1 have from 50 to 1,000 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 myotonic dystrophy type 1.

The size of the trinucleotide repeat expansion is associated with the severity of signs and symptoms. People with the classic features of myotonic dystrophy type 1, including muscle weakness and wasting beginning in adulthood, usually have between 100 and 1,000 CTG repeats in their cells. People born with the more severe, congenital form of myotonic dystrophy type 1 tend to have more than 1,000 CTG repeats in their cells. People with the mild form of the condition usually have between 50 and 150 CTG repeats in their cells.

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 do not develop myotonic dystrophy type 1, 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.

Other Names for This Gene

- DM protein kinase
- DM-kinase
- DM-PK
- DM1
- Dm15
- DM1PK
- 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

Tests Listed in the Genetic Testing Registry

- Tests of DMPK (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1760[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28DMPK%5BTIAB%5D%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; DMPK (https://omim.org/entry/605377)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1760)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DMPK[gene])

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Genomic Location

The *DMPK* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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