

## DNM2 gene

dynamin 2

### Normal Function

The *DNM2* gene provides instructions for making a protein called dynamin 2. Dynamin 2 is present in cells throughout the body. It is involved in endocytosis, which is a process that brings substances into the cell. During endocytosis, the cell membrane folds around a substance (such as a protein) outside the cell to form a sac-like structure called a vesicle. The vesicle is drawn into the cell and is pinched off from the cell membrane. Dynamin 2 is thought to play a key role in altering the cell membrane to form these vesicles.

Dynamin 2 is also involved in the cell's structural framework (cytoskeleton). The protein interacts with multiple parts of the cytoskeleton, including tube-like structures called microtubules and proteins called actin, which organize into filaments to provide structure. These parts of the cytoskeleton are involved in movement of molecules within the cells, cell shape, cell mobility, and attachment of cells to one another.

### Health Conditions Related to Genetic Changes

#### Centronuclear myopathy

At least 25 mutations in the *DNM2* gene have been found to cause centronuclear myopathy, a condition that is characterized by muscle weakness (myopathy) in the skeletal muscles, which are the muscles used for movement. Most of these mutations change single DNA building blocks (nucleotides) in regions of the gene known as exon 8, exon 11, and exon 16. These mutations lead to a change in the structure of dynamin 2. *DNM2* gene mutations that cause centronuclear myopathy are described as "gain-of-function" because they appear to enhance the activity of dynamin 2, affecting endocytosis and leading to disorganization of structures similar to microtubules, called transverse tubules (T tubules), which are found within the membrane of muscle fibers. The T tubules are necessary for normal muscle tensing (contractions) and relaxation. As a result of the *DNM2* gene mutations, the structure of muscle cells becomes abnormal and they cannot contract and relax normally, leading to the muscle weakness that is characteristic of centronuclear myopathy.

#### Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

## Other Names for This Gene

- CMT2M
- CMTDI1
- CMTDIB
- DI-CMTB
- DYN2
- DYN2\_HUMAN
- dynamin II
- DYNII

## Additional Information & Resources

## Tests Listed in the Genetic Testing Registry

- Tests of DNM2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1785\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1785[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DNM2%5BTIAB%5D%29+OR+%28dynamin+2%5BTIAB%5D%29%29+OR+%28%28DYN2%5BTIAB%5D%29+OR+%28dynamin+II%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5BDp%5D>)

## Catalog of Genes and Diseases from OMIM

- DYNAMIN 2; DNM2 (<https://omim.org/entry/602378>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1785>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DNM2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DNM2[gene]))

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## **Genomic Location**

The *DNM2* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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