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
Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 10,718,053 to 10,831,910 on chromosome 19 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Other Names for This Gene

• CMT2M
• CMTDI1
• CMTDIB
• DI-CMTB
• DYN2
• DYN2_HUMAN
• dynamin II
• DYNII

Additional Information & Resources

Educational Resources

• Annual Reviews Collection (2000): Dynamin and its Role in Membrane Fission
  https://www.ncbi.nlm.nih.gov/books/NBK2233/
  https://www.ncbi.nlm.nih.gov/books/NBK9831/

Clinical Information from GeneReviews

• Charcot-Marie-Tooth Hereditary Neuropathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1358
• DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy
  https://www.ncbi.nlm.nih.gov/books/NBK45014
Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- DYNAMIN 2
  http://omim.org/entry/602378

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DNM2.html
- ClinVar
- HGNC Gene Family: Pleckstrin homology domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/682
- HGNC Gene Symbol Report
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=38
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1785
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P50570

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301532
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17932957
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18394888

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26199319

• OMIM: DYNAMIN 2 
  http://omim.org/entry/602378

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17636067

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19130742

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25566070 
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4271577/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16775378

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20181480

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15180823

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15731758

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