NEFL gene
neurofilament light

Normal Function

The NEFL gene provides instructions for making the smallest protein component (the light subunit) of neurofilaments, which are essential for normal nerve cell function. Neurofilaments are assembled from light, medium, and heavy subunits. They form a structural framework that helps to define the shape and size of nerve cells. Cross-linking or bridging between neurofilaments maintains the diameter of the fiber, or axon, that extends from a nerve cell. Maintaining the proper axon diameter is essential for the transmission of nerve impulses.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

Researchers have identified more than 10 NEFL gene mutations that cause two forms of Charcot-Marie-Tooth disease known as type 1F and type 2E. Most NEFL gene mutations change single protein building blocks (amino acids) in the neurofilament light subunit. Other mutations delete or duplicate part of the NEFL gene, which alters the instructions for making the neurofilament light subunit. NEFL gene mutations probably alter the size or shape of neurofilament light subunits.

Altered neurofilament light subunits likely disrupt the assembly of neurofilaments or their transport to the axon. These disruptions may cause abnormalities in axons and impair the transmission of nerve impulses. Abnormalities in axons are a sign of type 2E Charcot-Marie-Tooth disease. In some cases, a reduced axon diameter may result in decreased nerve signal transmission speed, which is characteristic of type 1F Charcot-Marie-Tooth disease.
Chromosomal Location

Cytogenetic Location: 8p21.2, which is the short (p) arm of chromosome 8 at position 21.2

Molecular Location: base pairs 24,950,955 to 24,956,869 on chromosome 8 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT1F
- CMT2E
- neurofilament triplet L protein
- neurofilament, light polypeptide
- neurofilament, light polypeptide 68kDa
- NF68
- NFL
- NFL_HUMAN
- PPP1R110

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK21560/
  https://www.ncbi.nlm.nih.gov/books/NBK9834/
Clinical Information from GeneReviews

- Charcot-Marie-Tooth Hereditary Neuropathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1358
- Charcot-Marie-Tooth Neuropathy Type 2E/1F
  https://www.ncbi.nlm.nih.gov/books/NBK1187

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28NEFL%5BTIAB%5D%29+OR+%28NEFL%5BTIAB%5D%29+OR+%28NEFL+tri+prot%29+AND+English+AND+human
  last 1440 days

Catalog of Genes and Diseases from OMIM

- NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE
  http://omim.org/entry/162280

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_NEFL.html
- ClinVar
- HGNC Gene Family: Intermediate filaments Type IV
  https://www.genenames.org/cgi-bin/genefamilies/set/611
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits
  https://www.genenames.org/cgi-bin/genefamilies/set/694
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7739
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=8
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4747
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P07196
Sources for This Summary


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14685682

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17881652

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