NEB gene
nebulin

Normal Function

The *NEB* gene provides instructions for making a protein called nebulin. This protein plays an important role in skeletal muscles, which are muscles used for movement. Within skeletal muscle cells, nebulin is found in structures called sarcomeres. Sarcomeres are necessary for muscles to tense (contract). Nebulin is one of several proteins that interact to generate the mechanical force needed for muscle contraction.

Health Conditions Related to Genetic Changes

Nemaline myopathy

More than 60 mutations in the *NEB* gene have been found to cause nemaline myopathy. Some of these mutations change single protein building blocks (amino acids) in the nebulin protein or result in the production of an abnormally short protein. Other mutations insert or delete genetic material in the *NEB* gene. Most *NEB* gene mutations alter the structure or reduce the production of nebulin. A lack of functional nebulin impairs the muscle cells' ability to contract. Inefficient muscle contraction leads to muscle weakness and the other features of nemaline myopathy.

Chromosomal Location

Cytogenetic Location: 2q23.3, which is the long (q) arm of chromosome 2 at position 23.3

Molecular Location: base pairs 151,485,334 to 151,734,487 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- NEBU_HUMAN
- NEM2

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791
- Molecular Cell Biology (fourth edition, 2000): Titin and Nebulin Filaments Organize the Sarcomere
  https://www.ncbi.nlm.nih.gov/books/NBK21670/#A5209

Clinical Information from GeneReviews

- Nemaline Myopathy
  https://www.ncbi.nlm.nih.gov/books/NBK1288

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NEB%5BTIAB%5D%29+OR+%28nebulin%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+AND+human%5Bmh%5D+AND+\%22last+1800+days%22+AND+%22last+1800+days%22

Catalog of Genes and Diseases from OMIM

- NEBULIN
  http://omim.org/entry/161650

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_NEB.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=NEB%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4703
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P20929
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16917880

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19944167
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2856782/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11558787

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