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.

Other Names for This Gene

- NEBU_HUMAN
- NEM2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NEB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4703[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- NEBULIN; NEB (https://omim.org/entry/161650)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4703)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NEB[gene])

References

- Lehtokari VL, Pelin K, Sandbacka M, Ranta S, Donner K, Muntoni F, Sewry C, Angelini C, Bushby K, Van den Bergh P, Iannaccone S, Laing NG,Wallgren-Pettersson C. Identification of 45 novel mutations in the nebulin geneassociated with autosomal recessive nemaline myopathy. Hum Mutat. 2006Sep;27(9):946-56. doi: 10.1002/humu.20370. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16917880)
- Ottenheijm CA, Hooijman P, DeChene ET, Stienen GJ, Beggs AH, Granzier H. Altered myofilament function depresses force generation in patients withnebulin-based nemaline myopathy (NEM2). J Struct Biol. 2010 May;170(2):334-43.doi: 10.1016/j.jsb.2009.11.013. Epub 2009 Nov 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19944167) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2856782/)
- Ryan MM, Schnell C, Strickland CD, Shield LK, Morgan G, Iannaccone ST, LaingNG, Beggs AH, North KN. Nemaline myopathy: a clinical study of 143 cases. AnnNeurol. 2001 Sep;50(3):312-20. doi: 10.1002/ana.1080. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11558787)

Genomic Location

The *NEB* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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