SBF2 gene
SET binding factor 2

Normal Function
The SBF2 gene (also called MTMR13) provides instructions for making a protein called SET binding factor 2. The function of this protein is unknown, but it is probably involved in the development of specialized cells in the nervous system called Schwann cells. Schwann cells produce myelin, the protective substance that covers nerve cells and promotes the rapid transmission of nerve impulses. SET binding factor 2 probably also plays a role in the development of mesh-like canals (trabecular meshwork) that surround the colored part of the eye (the iris). The trabecular meshwork helps drain excess fluid from the eye.

Health Conditions Related to Genetic Changes
Charcot-Marie-Tooth disease
At least five SBF2 gene mutations have been identified in patients with a form of Charcot-Marie-Tooth disease known as type 4B2. Some of these mutations alter the structure of SET binding factor 2 by introducing a premature stop signal that results in an abnormally short protein. Other mutations lead to the production of a protein that is missing a critical segment. All of these mutations probably result in a nonfunctional protein. Although it is unclear how SBF2 gene mutations lead to this disorder, myelin production is probably disrupted. Irregular myelin structure (called outfolding) is a characteristic sign of type 4B2 Charcot-Marie-Tooth disease.

Individuals with this disorder may also experience a buildup of fluid pressure within the eye (glaucoma) beginning in childhood or adolescence. Researchers believe that the appearance of glaucoma depends on the type of SBF2 gene mutation. A mutation that causes complete loss of protein function interferes with the development of the eye’s trabecular meshwork, leading to impaired fluid drainage and glaucoma. Less severe mutations, which allow partial function of the SET binding factor 2 protein, do not cause glaucoma.
Chromosomal Location

Cytogenetic Location: 11p15.4, which is the short (p) arm of chromosome 11 at position 15.4

Molecular Location: base pairs 9,778,667 to 10,294,207 on chromosome 11 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene
- CMT4B2
- DENND7B
- KIAA1766
- MTMR13
- MTMRD_HUMAN

Additional Information & Resources

Educational Resources

GeneReviews

Scientific Articles on PubMed
- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28CMT4B2%5BTIAB%5D%29+OR+%28MTMR13%5BTIAB%5D%29+OR+%28SBF2%5BTIAB%5D%29+OR+%28SET+binding+factor+2%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+human%5Bmh%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D
OMIM

- SET-BINDING FACTOR 2
  http://omim.org/entry/607697

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SBF2.html

- ClinVar

- HGNC Gene Family: DENN/MADD domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/504

- HGNC Gene Family: Myotubularins
  https://www.genenames.org/cgi-bin/genefamilies/set/903

- HGNC Gene Family: Pleckstrin homology domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/682

- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2135

- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=24

- NCBI Gene

- UniProt
  http://www.uniprot.org/uniprot/Q86WG5

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12687498
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180267/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301641
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15477569

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16775368

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15304601

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18257469

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12554688

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

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