HSPB1 gene

heat shock protein family B (small) member 1

Normal Function

The *HSPB1* gene provides instructions for making a protein called heat shock protein beta-1 (also called heat shock protein 27). This protein is a member of the heat shock protein family, which helps protect cells under adverse conditions such as infection, inflammation, exposure to toxins, elevated temperature, injury, and disease. Heat shock proteins block signals that lead to programmed cell death. In addition, they appear to be involved in activities such as cell movement (motility), stabilizing the cell's structural framework (the cytoskeleton), folding and stabilizing newly produced proteins, and repairing damaged proteins. Heat shock proteins also appear to play a role in the tensing of muscle fibers (muscle contraction).

Heat shock protein beta-1 is found in cells throughout the body and is particularly abundant in nerve and muscle cells. In nerve cells, this protein helps to organize a network of molecular threads called neurofilaments that maintain the diameter of specialized extensions called axons. Maintaining proper axon diameter is essential for the efficient transmission of nerve impulses. Although it is thought to play a role in muscle contraction, the specific function of heat shock protein beta-1 in muscle cells is unclear.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

Distal hereditary motor neuropathy, type II

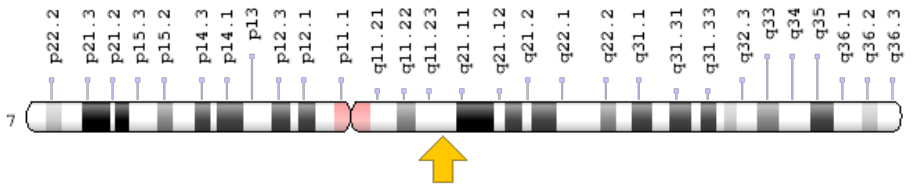
Researchers have identified at least 14 *HSPB1* gene mutations that cause a condition called distal hereditary motor neuropathy, type II, which is characterized by progressive weakness, primarily in the feet and legs.

HSPB1 gene mutations that cause distal hereditary motor neuropathy, type II change single protein building blocks (amino acids) in heat shock protein beta-1. Studies suggest that the altered protein may be more likely to form clusters (aggregates) and block the transport of substances that are essential for the proper function of nerve axons, leading to the signs and symptoms of distal hereditary motor neuropathy, type II.

Chromosomal Location

Cytogenetic Location: 7q11.23, which is the long (q) arm of chromosome 7 at position 11.23

Molecular Location: base pairs 76,302,673 to 76,304,292 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT2F
- heat shock 27kDa protein 1
- heat shock protein beta-1
- HS.76067
- Hsp25
- HSP27
- HSP28
- HSPB1_HUMAN
- SRP27
- stress-responsive protein 27

Additional Information & Resources

Clinical Information from GeneReviews

- Charcot-Marie-Tooth Hereditary Neuropathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1358

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28HSPB1%5BTIAB%5D%29+OR+%28HSP27%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- HEAT-SHOCK 27-KD PROTEIN 1
  http://omim.org/entry/602195

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/HSPB1ID40880ch7q11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HSPB1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:5246
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3315
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3315
- UniProt
  https://www.uniprot.org/uniprot/P04792

Sources for This Summary

- Ackerley S, James PA, Kalli A, French S, Davies KE, Talbot K. A mutation in the small heat-shock protein HSPB1 leading to distal hereditary motor neuronopathy disrupts neurofilament assembly and the axonal transport of specific cellular cargoes. Hum Mol Genet. 2006 Jan 15;15(2):347-54. Epub 2005 Dec 20.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16368711
- Bird TD. Charcot-Marie-Tooth Hereditary Neuropathy Overview. 1998 Sep 28 [updated 2016 Sep 1]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from http://www.ncbi.nlm.nih.gov/books/NBK1358/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301532
- Datskevich PN, Nefedova VV, Sudnitsyna MV, Gusev NB. Mutations of small heat shock proteins and human congenital diseases. Biochemistry (Mosc). 2012 Dec;77(13):1500-14. doi: 10.1134/S0006297912130081. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23379525
- Dierick I, Irobi J, De Jonghe P, Timmerman V. Small heat shock proteins in inherited peripheral neuropathies. Ann Med. 2005;37(6):413-22. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16203614

- Drew AP, Blair IP, Nicholson GA. Molecular genetics and mechanisms of disease in distal hereditary motor neuropathies: insights directing future genetic studies. Curr Mol Med. 2011 Nov; 11(8):650-65. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21902652
- Evgrafov OV, Mersiyanova I, Irobi J, Van Den Bosch L, Dierick I, Leung CL, Schagina O, Verpoorten N, Van Impe K, Fedotov V, Dadali E, Auer-Grumbach M, Windpassinger C, Wagner K, Mitrovic Z, Hilton-Jones D, Talbot K, Martin JJ, Vasserman N, Tverskaya S, Polyakov A, Liem RK, Gettemans J, Robberecht W, De Jonghe P, Timmerman V. Mutant small heat-shock protein 27 causes axonal Charcot-Marie-Tooth disease and distal hereditary motor neuropathy. Nat Genet. 2004 Jun;36(6):602-6. Epub 2004 May 2.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15122254
- OMIM: HEAT-SHOCK 27-KD PROTEIN 1
  http://omim.org/entry/602195
- Houlden H, Laura M, Wavrant-De Vrièze F, Blake J, Wood N, Reilly MM. Mutations in the HSP27 (HSPB1) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. Neurology. 2008 Nov 18;71(21):1660-8. doi: 10.1212/01.wnl.0000319696.14225.67. Epub 2008 Oct 1.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18832141
- Kampinga HH, Garrido C. HSPBs: small proteins with big implications in human disease. Int J Biochem Cell Biol. 2012 Oct;44(10):1706-10. doi: 10.1016/j.biocel.2012.06.005. Epub 2012 Jun 19. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22721753
- Nefedova VV, Muranova LK, Sudnitsyna MV, Ryzhavskaya AS, Gusev NB. Small Heat Shock Proteins and Distal Hereditary Neuropathies. Biochemistry (Mosc). 2015 Dec;80(13):1734-47. doi: 10.1134/S000629791513009X. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26878578
- Singh MK, Sharma B, Tiwari PK. The small heat shock protein Hsp27: Present understanding and future prospects. J Therm Biol. 2017 Oct;69:149-154. doi: 10.1016/j.jtherbio.2017.06.004. Epub 2017 Jun 7. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/29037376
- Stetler RA, Gao Y, Signore AP, Cao G, Chen J. HSP27: mechanisms of cellular protection against neuronal injury. Curr Mol Med. 2009 Sep;9(7):863-72. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19860665
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2775412/
- Wettstein G, Bellaye PS, Micheau O, Bonniaud P. Small heat shock proteins and the cytoskeleton: an essential interplay for cell integrity? Int J Biochem Cell Biol. 2012 Oct;44(10):1680-6. doi: 10.1016/j.biocel.2012.05.024. Epub 2012 Jun 7. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22683760

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