

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

MedlinePlus Genetics provides information about 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.

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

Tests Listed in the Genetic Testing Registry

- Tests of HSPB1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3315\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3315[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

Catalog of Genes and Diseases from OMIM

- HEAT-SHOCK 27-KD PROTEIN 1; HSPB1 (<https://omim.org/entry/602195>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3315>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HSPB1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HSPB1[gene]))

References

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Genomic Location

The *HSPB1* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

Last updated January 1, 2010