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. This protein is a member of the heat shock protein family, which helps protect cells that are under stress from factors such as infection, inflammation, exposure to toxins, elevated temperature, injury, and disease. Heat shock proteins block signals that lead cells to self-destruct (undergo apoptosis). In addition, they appear to be involved in cell movement, stabilizing the cell's structural framework (the cytoskeleton), folding and stabilizing newly produced proteins, and repairing damaged proteins. Heat shock proteins also seem to play a role in the tensing of muscle fibers (muscle contraction).

Heat shock protein beta-1 is found in cells throughout the body, where it interacts with other proteins and primarily helps fold newly produced proteins and refold damaged proteins. In nerve cells (neurons), heat shock protein beta-1 helps to organize a network of molecular threads called neurofilaments that maintain the structure of a part of the neuron called the axon. This allows neurons to transmit nerve impulses efficiently.

Health Conditions Related to Genetic Changes

Distal hereditary motor neuropathy, type II

Variants (also called mutations) in the *HSPB1* gene have been found to cause a condition called distal hereditary motor neuropathy, type II. This condition is characterized by damage to specialized neurons in the brain and spinal cord that control muscle movement (motor neurons). Damage to motor neurons leads to progressive weakness and loss of muscle tissue (atrophy) in the feet and legs. *HSPB1* gene variants are the most common cause of distal hereditary motor neuropathy, type II.

The *HSPB1* gene variants that cause distal hereditary motor neuropathy, type II typically lead to changes in 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). These aggregates can build up and impair the function of cells, particularly motor neurons. In addition, motor neurons that do not have a functional version of heat shock protein beta-1 are vulnerable to stress and cell damage. As a result, these cells are more likely to die off over time, leading to the signs and symptoms of distal hereditary motor neuropathy, type II.

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Other Names for This Gene

- heat shock protein beta-1
- 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])

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)

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])

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Genomic Location

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

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