HSPB8 gene

heat shock protein family B (small) member 8

Normal Function

The *HSPB8* gene provides instructions for making a protein called heat shock protein beta-8 (also called heat shock protein 22). 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-8 is found in cells throughout the body and is particularly abundant in nerve cells. While its function is not well understood, it seems to interact with a related protein called heat shock protein beta-1, produced from the *HSPB1* gene. In nerve cells, heat shock protein beta-1 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. The specific role that heat shock protein beta-8 plays in axons 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 five *HSPB8* gene mutations that cause a condition called distal hereditary motor neuropathy, type II. This disorder is characterized by progressive weakness, primarily in the feet and legs.

It is unclear how HSPB8 gene mutations lead to the signs and symptoms of distal hereditary motor neuropathy, type II. Research suggests that the altered heat shock protein beta-8 interacts more strongly with heat shock protein beta-1 and is more likely

to form clumps (aggregates). The aggregates may 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

- CMT2L
- DHMN2
- E2-induced gene 1
- E2IG1
- H11
- heat shock 22kDa protein 8
- heat shock 27kDa protein 8
- heat shock protein beta-8
- HMN2
- HMN2A
- HSP22
- HspB8
- HSPB8_HUMAN
- protein kinase H11
- small stress protein-like protein HSP22

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HSPB8 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26353[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28HSPB8%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- HEAT-SHOCK 22-KD PROTEIN 8; HSPB8 (https://omim.org/entry/608014)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/26353)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HSPB8[gene])

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Genomic Location

The *HSPB8* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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