THAP1 gene

THAP domain containing 1

Normal Function

The *THAP1* gene provides instructions for making a protein that is a transcription factor, which means that it attaches (binds) to specific regions of DNA and regulates the activity of other genes. Through this function, it is thought to help control several processes in the body, including the growth and division (proliferation) of endothelial cells, which line the inside surface of blood vessels and other circulatory system structures called lymphatic vessels. The THAP1 protein also plays a role in the self-destruction of cells that are no longer needed (apoptosis).

Health Conditions Related to Genetic Changes

Dystonia 6

More than 70 *THAP1* gene mutations have been identified in people with dystonia 6. Dystonia 6 is one of many forms of dystonia, which is a group of conditions characterized by involuntary movements, twisting (torsion) and tensing of various muscles, and unusual positioning of affected body parts.

Most of the *THAP1* gene mutations that cause dystonia 6 change single protein building blocks (amino acids) in the THAP1 protein or result in a premature stop signal that leads to an abnormally short protein. Studies indicate that many of the mutations affect the stability of the THAP1 protein, reducing the amount of functional THAP1 protein available for DNA binding. Others may impair the protein's ability to bind with the correct regions of DNA. Problems with DNA binding likely disrupt the proper regulation of gene activity, leading to the signs and symptoms of dystonia 6.

A particular *THAP1* gene mutation is specific to a Mennonite population in the Midwestern United States in which dystonia 6 was first described. This mutation changes the DNA sequence in a region of the gene known as exon 2. Some researchers use the term DYT6 dystonia to refer to dystonia caused by this particular mutation, and the broader term THAP1 dystonia to refer to dystonia caused by any *THAP1* gene mutation. In general, mutations affecting the region of the THAP1 protein that binds to DNA, including the mutation found in the Mennonite population, tend to result in more severe signs and symptoms than mutations affecting other regions of the protein.

Other Names for This Gene

- 4833431A01Rik
- DYT6
- FLJ10477
- nuclear proapoptotic factor
- THAP domain containing, apoptosis associated protein 1
- THAP domain protein 1
- THAP domain-containing protein 1
- THAP1_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of THAP1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55145[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28THAP1%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

- THAP DOMAIN-CONTAINING PROTEIN 1; THAP1 (https://omim.org/entry/609520)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55145)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=THAP1[gene])

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Genomic Location

The *THAP1* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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