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.

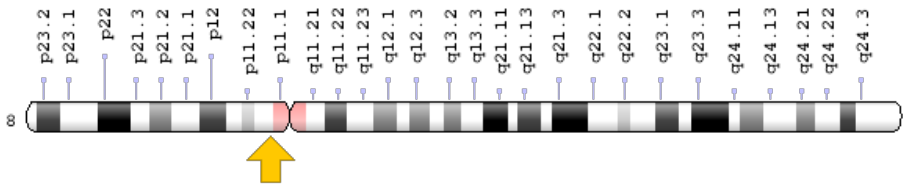
Most of the *THAP1* gene mutations that cause dystonia 6 change single protein building blocks (amino acids) in the THAP1 protein or result in a premature stop signal that leads to an abnormally short protein. Studies indicate that many of the mutations affect the stability of the THAP1 protein, reducing 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.

Chromosomal Location

Cytogenetic Location: 8p11.21, which is the short (p) arm of chromosome 8 at position 11.21

Molecular Location: base pairs 42,836,674 to 42,843,331 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?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

OMIM

- THAP DOMAIN-CONTAINING PROTEIN 1 http://omim.org/entry/609520

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_THAP1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=THAP1%5Bgene%5D
- HGNC Gene Family: THAP domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/65
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20856
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/55145
- UniProt
  http://www.uniprot.org/uniprot/Q9NVV9
- Universal Mutation Database: THAP1 Mutations Database
  http://www.umd.be/THAP1/

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