TBP gene
TATA-box binding protein

Normal Function

The TBP gene provides instructions for making a protein called the TATA box binding protein. This protein is active in cells and tissues throughout the body, where it plays an essential role in regulating the activity of most genes.

The TATA box binding protein attaches (binds) to a particular sequence of DNA known as the TATA box. This sequence occurs in a regulatory region of DNA near the beginning of many genes. Once the protein is attached to the TATA box near a gene, it acts as a landmark to indicate where other enzymes should start reading the gene. The process of reading a gene’s DNA and transferring the information to a similar molecule called mRNA is known as transcription.

One region of the TBP gene contains a particular DNA segment known as a CAG/CAA trinucleotide repeat. This segment is made up of a series of three DNA building blocks (nucleotides) that appear multiple times in a row. Normally, the CAG/CAA segment is repeated 25 to 42 times within the gene.

Health Conditions Related to Genetic Changes

Huntington disease-like syndrome

A particular type of mutation in the TBP gene has been found to cause a progressive brain disorder known as Huntington disease-like 4 (HDL4) or spinocerebellar ataxia type 17 (SCA17). The features of this disorder vary widely among affected individuals. The condition was first described as HDL4 in people whose signs and symptoms closely resembled those of Huntington disease, including uncontrolled movements, emotional problems, and loss of thinking ability. The disorder is now more commonly known as SCA17 because difficulty coordinating movements (ataxia) and other movement problems are the most frequent signs and symptoms. It is unknown why some people with TBP mutations have a disorder resembling Huntington disease, while others have more prominent ataxia.

The mutation associated with HDL4/SCA17 increases the size of the CAG/CAA trinucleotide repeat in the TBP gene. People with this condition have 43 to 66 CAG/CAA repeats. People with 43 to 48 CAG/CAA repeats may or may not have signs and symptoms, while people with 49 or more repeats almost always develop the disorder.

An increased number of CAG/CAA repeats in the TBP gene leads to the production of an abnormally long version of the TATA box binding protein. The abnormal protein builds up in nerve cells (neurons) in the brain and disrupts the normal functions of
these cells. The dysfunction and eventual death of neurons in certain areas of the brain underlie the signs and symptoms of HDL4/SCA17. Because the TBP gene is active throughout the body, it is unclear why the effects of a mutation in this gene are limited to the brain.

**Chromosomal Location**

Cytogenetic Location: 6q27, which is the long (q) arm of chromosome 6 at position 27

Molecular Location: base pairs 170,554,333 to 170,572,870 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- CCG1 Protein
- CCGS
- Cell Cycle Gene 1 Protein
- DYT3 protein, human
- GTF2D
- GTF2D1
- RNA Polymerase II TATA-Binding Protein
- RNA Polymerase IIA 250kD
- SCA17
- TAF(II)250
- TAF1 RNA Polymerase II TATA Box Binding Protein
- TAF2A
- TAFII250
- TATA-Binding Protein
- TATA box binding protein
- TATA-Box Factor
- TATA Sequence-Binding Protein
- TBP_HUMAN
- TF2D
- TFIID
- Transcription Factor IID
- Transcription Factor TBP
- Transcription Initiation Factor TFIID 250 kDa Subunit

**Additional Information & Resources**

**Educational Resources**
- Biochemistry (fifth edition, 2002): The TATA-Box-Binding Protein Initiates the Assembly of the Active Transcription Complex
  https://www.ncbi.nlm.nih.gov/books/NBK22433/#A3988
- Protein Data Bank Molecule of the Month: TATA-Binding Protein
  http://pdb101.rcsb.org/motm/67

**Clinical Information from GeneReviews**
- Spinocerebellar Ataxia Type 17
  https://www.ncbi.nlm.nih.gov/books/NBK1438

**Scientific Articles on PubMed**
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TATA+box+binding+protein%5BMAJR%5D%29+OR+%28SCA17%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

**Catalog of Genes and Diseases from OMIM**
- TATA BOX-BINDING PROTEIN
  http://omim.org/entry/600075

**Research Resources**
- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TBP.html
- ClinVar
- HGNC Gene Symbol Report
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14985389
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735701/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18043721

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17474109

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12953269

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17101913

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11313753

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15916858

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