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's disease-like syndrome

A particular type of variant (also called a mutation) in the *TBP* gene has been found to cause a progressive brain disorder known as Huntington's 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's 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* gene variants have a disorder resembling Huntington's disease, while others have more prominent ataxia.

The variants 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 variant in this gene are limited to the brain.

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 box binding protein
- TATA Sequence-Binding Protein
- TATA-Binding Protein
- TATA-Box Factor
- TBP_HUMAN
- TF2D
- TFIID
- Transcription Factor IID
- Transcription Factor TBP
- Transcription Initiation Factor TFIID 250 kDa Subunit

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TBP (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6908[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; TBP (https://omim.org/entry/600075)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6908)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TBP[gene])

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Genomic Location

The *TBP* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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