TAF1 gene

TATA-box binding protein associated factor 1

Normal Function

The *TAF1* gene provides instructions for making part of a protein called transcription factor IID (TFIID). This protein is active in cells and tissues throughout the body, where it attaches (binds) to DNA. Transcription factor IID plays an essential role in regulating the activity of most genes.

The *TAF1* gene is part of a complex region of DNA known as the *TAF1/DYT3* multiple transcript system. This region consists of short stretches of DNA from the *TAF1* gene plus some extra segments of genetic material near the gene. These stretches of DNA can be combined in different ways to create various sets of instructions for making proteins. Researchers believe that some of these variations are critical for the normal function of nerve cells (neurons) in the brain.

Health Conditions Related to Genetic Changes

X-linked dystonia-parkinsonism

Several changes in the *TAF1/DYT3* multiple transcript system have been identified in people with X-linked dystonia-parkinsonism. This condition affects movement and has been found only in people of Filipino descent.Some gene changes alter single DNA building blocks (nucleotides); these changes are described as disease-specific single-nucleotide changes (DSCs). Another genetic change deletes a small number of nucleotides from the gene. Researchers are uncertain how these changes are related to the movement abnormalities characteristic of the disease.

X-linked dystonia-parkinsonism may also be related to an extra segment of DNA in the *TAF1/DYT3* multiple transcript system. The extra segment results from the insertion of a retrotransposon, which is a small piece of DNA that can move around to different positions in a cell's genetic material. When a retrotransposon inserts itself in or a near a gene, it can disrupt the gene's function. In this case, the retrotransposon insertion probably interferes with the normal function of the *TAF1/DYT3* multiple transcript system.

Researchers suspect that changes in the *TAF1/DYT3* multiple transcript system disrupt the regulation of critical genes in neurons. This defect leads to the eventual death of

these cells, particularly in areas of the brain called the caudate nucleus and putamen. These regions are critical for normal movement, learning, and memory. It is unclear why the effects of changes in the *TAF1/DYT3* multiple transcript system appear to be limited to dystonia and parkinsonism.

Other Names for This Gene

- BA2R
- CCG1
- CCGS
- Cell cycle gene 1 protein
- cell cycle, G1 phase defect
- complementation of cell cycle block, G1-to-S
- DYT3
- DYT3/TAF1
- KAT4
- N-TAF1
- NSCL2
- P250
- TAF(II)250
- TAF1 RNA polymerase II, TATA box binding protein (TBP)-associated factor, 250kDa
- TAF1_HUMAN
- TAF2A
- TAFII-250
- TAFII250
- TATA box binding protein associated factor 1
- TATA box-binding protein-associated factor 1
- TATA box-binding protein-associated factor 2A
- TBP-associated factor 1
- TBP-associated factor 250 kDa
- TBP-associated factor, RNA polymerase II, 250-kD
- transcription factor TFIID p250 polypeptide
- Transcription initiation factor TFIID 250 kDa subunit
- Transcription initiation factor TFIID subunit 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TAF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6872[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TAF1%5BTIAB%5D%29+OR+%28DYT3%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TAF1 RNA POLYMERASE II, TATA BOX-BINDING PROTEIN-ASSOCIATED FACTOR, 250-KD; TAF1 (https://omim.org/entry/313650)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6872)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TAF1[gene])

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Genomic Location

The *TAF1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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