GTF2I gene

general transcription factor IIi

Normal Function

The *GTF2I* gene provides instructions for making two proteins, TFII-I and BAP-135. TFII-I attaches (binds) to specific areas of DNA and helps regulate the activity of other genes. Based on this role, TFII-I is called a transcription factor. This protein is active in the brain and many other tissues in the body. Studies suggest that the TFII-I protein is involved in coordinating cell growth and division, and it may also play a role in controlling the flow of calcium into cells. Studies show it may be important in a process called myelination, which is the formation of the protective coating around nerve cells called the myelin sheath. This coating insulates nerve cells and promotes the rapid transmission of nerve impulses.

Less is known about BAP-135, the other protein produced from the *GTF2I* gene. The protein is active in B cells, which are a specialized type of white blood cell that protects the body against infection. When a B cell senses a foreign substance (such as a virus), it triggers a series of chemical reactions that instruct the cell to mature, divide, and produce specific proteins called antibodies to fight the infection. The BAP-135 protein is turned on as part of this series of chemical reactions.

Health Conditions Related to Genetic Changes

7q11.23 duplication syndrome

The *GTF2I* gene is located in a region of chromosome 7 that is duplicated in people with 7q11.23 duplication syndrome. As a result of this duplication, people with 7q11.23 duplication syndrome have an extra copy of the *GTF2I* gene and several other genes in each cell. 7q11.23 duplication syndrome can cause a variety of neurological and behavioral problems as well as other abnormalities.

Behavioral problems associated with 7q11.23 duplication syndrome include anxiety disorders (such as social phobias and selective mutism, which is an inability to speak in certain circumstances), attention-deficit/hyperactivity disorder (ADHD), physical aggression, excessively defiant behavior (oppositional disorder), and autistic behaviors that affect communication and social interaction. Studies suggest that an extra copy of the *GTF2I* gene may be associated with some of the behavioral features of 7q11.23 duplication syndrome, but the mechanism of this effect is unclear. Affected individuals

do not appear to have immune abnormalities related to this disorder.

Williams syndrome

The *GTF2I* gene is located in a region of chromosome 7 that is missing (deleted) in people with Williams syndrome, which is a developmental disorder characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems.As a result of the deletion, people with Williams syndrome are missing one copy of the *GTF2I* gene in each cell.

Studies suggest that the loss of this gene is partly responsible for intellectual disability in people with Williams syndrome. Loss of this gene may also contribute to behavioral differences, such as increased sociability and anxiety-related behaviors, that are seen in this disorder. Studies show that some affected individuals have less myelin than normal in some parts of their brains, and researchers suspect reduced myelination may contribute to the behavioral features. Researchers are investigating how changes in this gene may be related to these and other specific features of Williams syndrome. People with Williams syndrome do not appear to have immune abnormalities related to the condition.

Other Names for This Gene

- BAP-135
- BAP135
- Bruton tyrosine kinase-associated protein 135
- BTK-associated protein, 135kD
- BTKAP1
- DIWS
- GTF2I_HUMAN
- IB291
- SPIN
- TFII-I
- WBSCR6

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GTF2I (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2969[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28GTF2I%5BTIAB%5D%29+OR+%28%28BAP-135%5BTIAB%5D%29+OR+%28TFII-I%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GENERAL TRANSCRIPTION FACTOR II-I; GTF2I (https://omim.org/entry/601679)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2969)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GTF2I[gene])

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Genomic Location

The *GTF2I* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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