# GTF2I Gene

Subjects: Genetics & Heredity
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General transcription factor IIi

Keywords: genes

## 1. Introduction

The *GTF2I* gene provides instructions for making two proteins, BAP-135 and TFII-I. BAP-135 is involved in normal immune system function. It 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 activated as part of this series of chemical reactions; it transmits chemical signals that allow B cells to respond to potentially harmful invaders.

TFII-I, the other protein produced from the *GTF2I* gene, 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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. Despite the role of the *GTF2I* gene in immune function, affected individuals do not appear to have immune abnormalities related to this disorder.

### 2.2. Williams syndrome

The *GTF2I* gene is located in a region of chromosome 7 that is deleted in people with Williams syndrome. As a result of this deletion, people with this condition 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 dental abnormalities and the characteristic problems with visual-spatial tasks, such as writing and drawing, that are seen in this disorder. Researchers are investigating how a deletion involving this gene may be related to these specific features of Williams syndrome.

## 3. 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

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