

## **GTF2IRD1 gene**

GTF2I repeat domain containing 1

### **Normal Function**

The *GTF2IRD1* gene provides instructions for making a protein that regulates the activity of many other genes. This protein probably interacts with specific regions of DNA and with other proteins to turn genes on or off. Based on this role, the GTF2IRD1 protein is called a transcription factor.

Although its exact function is unknown, the *GTF2IRD1* gene is active in many of the body's tissues. It appears to be particularly important for gene regulation in the brain and in muscles used for movement (skeletal muscles). Studies suggest that this gene also plays a role in the development of tissues in the head and face (craniofacial development).

### **Health Conditions Related to Genetic Changes**

#### Williams syndrome

The *GTF2IRD1* gene is located in a region of chromosome 7 that is 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 *GTF2IRD1* gene in each cell. Studies suggest that the loss of this gene may contribute to some of the characteristic features of Williams syndrome, including the distinctive facial features, personality characteristics such as sociability, and problems with visual-spatial tasks such as writing and drawing. Researchers are investigating how a deletion of this gene may be related to these specific features.

### **Other Names for This Gene**

- CREAM1
- general transcription factor 3
- GT2D1\_HUMAN
- GTF2I repeat domain-containing 1
- GTF3

- hMusTRD1alpha1
- muscle TFII-I repeat domain-containing protein 1 alpha 1
- MusTRD1
- RBAP2
- WBSCR11

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of GTF2IRD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9569\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9569[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28GTF2IRD1%5BTIAB%5D%29+OR+%28%28MUSTRD1%5BTIAB%5D%29+OR+%28WBSCR11%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+d+ays%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- GTF2I REPEAT DOMAIN-CONTAINING PROTEIN 1; GTF2IRD1 (<https://omim.org/entry/604318>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9569>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GTF2IRD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GTF2IRD1[gene]))

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## Genomic Location

The *GTF2IRD1* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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