# 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

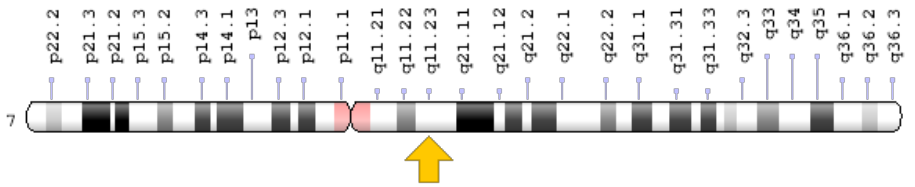
Williams syndrome

The *GTF2IRD1* gene is located in a region of chromosome 7 that is deleted in people with Williams syndrome. As a result of this deletion, people with this condition 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, dental abnormalities, 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.

## Chromosomal Location

Cytogenetic Location: 7q11.23, which is the long (q) arm of chromosome 7 at position 11.23

Molecular Location: base pairs 74,453,906 to 74,603,070 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)




Credit: Genome Decoration Page/NCBI

## 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

Educational Resources

- An Introduction to Genetic Analysis (seventh edition, 1996): Transcription: an overview of gene regulation in eukaryotes
  https://www.ncbi.nlm.nih.gov/books/NBK21780/

Clinical Information from GeneReviews

- Williams Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1249

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- GTF2I REPEAT DOMAIN-CONTAINING PROTEIN 1
  http://omim.org/entry/604318

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GTF2IRD1.html
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4661
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9569
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9569
- UniProt
  https://www.uniprot.org/uniprot/Q9UHL9

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