



## TBX1 gene

T-box 1

### Normal Function

The *TBX1* gene provides instructions for making a protein called T-box 1. Genes in the T-box family play important roles in the formation of tissues and organs during embryonic development. To carry out these roles, proteins produced from these genes bind to specific areas of DNA. The proteins attach to critical regions near genes and help control the activity of those genes. T-box proteins are called transcription factors on the basis of this action.

The T-box 1 protein appears to be necessary for the normal development of muscles and bones of the face and neck, large arteries that carry blood out of the heart, structures in the ear, and glands such as the thymus and parathyroid. Although the T-box 1 protein acts as a transcription factor, researchers have not determined which genes are regulated by this protein.

### Health Conditions Related to Genetic Changes

#### 22q11.2 deletion syndrome

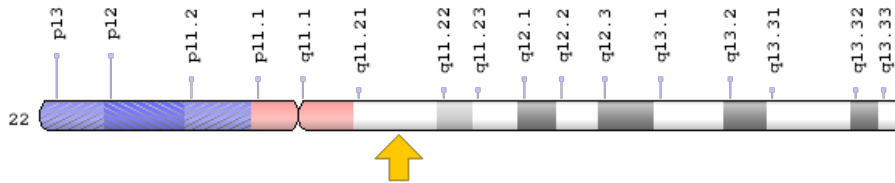
Most cases of 22q11.2 deletion syndrome are caused by a deletion of a small piece of chromosome 22. This region of the chromosome contains 30 to 40 genes, including the *TBX1* gene. In a small number of affected individuals without a chromosome 22 deletion, mutations in the *TBX1* gene are thought to be responsible for the characteristic signs and symptoms of the syndrome. The identified mutations include changes in single DNA building blocks (base pairs) in the *TBX1* gene and deletions of a small amount of genetic material from the gene. Some of these mutations reduce the amount of T-box 1 protein that is produced in cells, while other mutations alter the protein's function. These genetic changes likely affect the ability of the T-box 1 protein to bind to DNA and regulate the activity of other genes.

Researchers believe that changes in the *TBX1* gene, due to either a mutation in the gene or a deletion of part of chromosome 22, are responsible for many of the features of 22q11.2 deletion syndrome. Specifically, a reduction in the amount of T-box 1 or changes in the protein's normal function are associated with heart defects, an opening in the roof of the mouth (a cleft palate), distinctive facial features, hearing loss, and low calcium levels. Some studies suggest that a loss of the *TBX1* gene may also be associated with behavioral problems in affected individuals.

## Chromosomal Location

Cytogenetic Location: 22q11.21, which is the long (q) arm of chromosome 22 at position 11.21

Molecular Location: base pairs 19,756,703 to 19,783,593 on chromosome 22 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CAFS
- CTHM
- DGCR
- DGS
- DORV
- TBX1\_HUMAN
- TBX1C
- Testis-specific T-box protein
- TGA
- VCFS

## Additional Information & Resources

### Clinical Information from GeneReviews

- 22q11.2 Deletion Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1523>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TBX1%5BTIAB%5D%29+OR+%28T-box+1%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+1800+days%22%5Bdp%5D>

## Catalog of Genes and Diseases from OMIM

- T-BOX 1  
<http://omim.org/entry/602054>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_TBX1.html](http://atlasgeneticsoncology.org/Genes/GC_TBX1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=TBX1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:11592](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11592)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:6899>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6899>
- UniProt  
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