



ETV6 gene

ETS variant 6

Normal Function

The *ETV6* gene provides instructions for producing a protein that functions as a transcription factor, which means that it attaches (binds) to specific regions of DNA and controls the activity of certain genes. The ETV6 protein is found in the nucleus of cells throughout the body, where it turns off (represses) gene activity. It plays a key role in development before birth and in regulating blood cell formation.

Health Conditions Related to Genetic Changes

PDGFRB-associated chronic eosinophilic leukemia

PDGFRB-associated chronic eosinophilic leukemia, a type of cancer of blood-forming cells, can be caused by a genetic rearrangement known as a translocation that brings together part of the *ETV6* gene and part of another gene called *PDGFRB*, creating the *ETV6-PDGFRB* fusion gene. The translocation that leads to the *ETV6-PDGFRB* fusion gene is a somatic mutation, which is acquired during a person's lifetime and occurs initially in a single cell. This cell continues to grow and divide, producing a group of cells with the same mutation (a clonal population).

The protein produced from the *ETV6-PDGFRB* fusion gene, called ETV6/PDGFR β , functions differently than the proteins normally produced from the individual genes. Unlike the normal PDGFR β protein, the fusion protein is always active, which means certain cell signaling pathways are constantly turned on. The fusion protein is unable to repress gene activity regulated by the normal ETV6 protein, so gene activity is increased. The overactive signaling pathways and abnormal gene activity increase the proliferation and survival of cells. When the *ETV6-PDGFRB* fusion gene mutation occurs in cells that develop into blood cells, the growth of white blood cells called eosinophils (and occasionally other white blood cells, such as neutrophils and mast cells) is poorly controlled, leading to *PDGFRB*-associated chronic eosinophilic leukemia. It is unclear why eosinophils are preferentially affected by this genetic change.

Other cancers

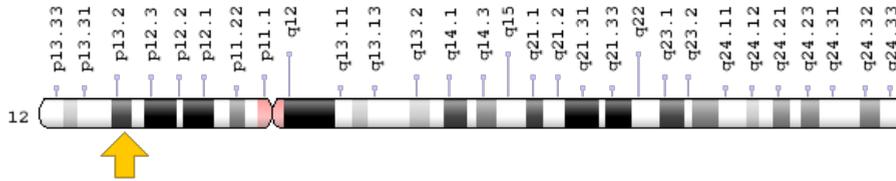
Translocations involving the *ETV6* gene and more than 30 other genes have been found to cause different types of leukemia, including acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL), and a bone marrow disease called myelodysplastic syndrome (MDS). Depending on the gene fused with *ETV6*, a number of mechanisms can cause these conditions, such as impaired regulation of

gene activity, abnormal signaling, or loss of normal gene function. The *ETV6* gene translocations that cause these diseases are somatic mutations and are not inherited.

Chromosomal Location

Cytogenetic Location: 12p13.2, which is the short (p) arm of chromosome 12 at position 13.2

Molecular Location: base pairs 11,649,601 to 11,895,402 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ETS-related protein Tel1
- ETS translocation variant 6
- ets variant 6
- ets variant gene 6 (TEL oncogene)
- ETV6_HUMAN
- TEL
- TEL1 oncogene
- transcription factor ETV6

Additional Information & Resources

Educational Resources

- Holland-Frei Cancer Medicine (sixth edition, 2003): Chromosomal Rearrangements <https://www.ncbi.nlm.nih.gov/books/NBK12538/#A1403>
- Holland-Frei Cancer Medicine (sixth edition, 2003): Transcription Factors <https://www.ncbi.nlm.nih.gov/books/NBK13714/#A1394>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ETV6%5BTIAB%5D%29+OR+%28TEL%5BTI%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

- ETS VARIANT GENE 6
<http://omim.org/entry/600618>
- LEUKEMIA, ACUTE MYELOID
<http://omim.org/entry/601626>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/ETV6ID38.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ETV6%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3495
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:2120>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2120>
- UniProt
<https://www.uniprot.org/uniprot/P41212>

Sources for This Summary

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<http://omim.org/entry/600618>
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