# RUNX1 gene

runt related transcription factor 1

## Normal Function

The *RUNX1* gene provides instructions for making a protein called runt-related transcription factor 1 (RUNX1). Like other transcription factors, the RUNX1 protein attaches (binds) to specific regions of DNA and helps control the activity of particular genes. This protein interacts with another protein called core binding factor beta or CBFβ (produced from the *CBFB* gene), which helps RUNX1 bind to DNA and prevents it from being broken down. Together, these proteins form one version of a complex known as core binding factor (CBF). The RUNX1 protein turns on (activates) genes that help control the development of blood cells (hematopoiesis). In particular, it plays an important role in development of hematopoietic stem cells, early blood cells that have the potential to develop into all types of mature blood cells such as white blood cells, red blood cells, and cell fragments called platelets.

## Health Conditions Related to Genetic Changes

Core binding factor acute myeloid leukemia

A rearrangement (translocation) of genetic material involving the *RUNX1* gene is found in approximately 7 percent of individuals with a form of blood cancer known as acute myeloid leukemia (AML). The translocation, written as t(8;21), combines genetic information from chromosome 21 and chromosome 8, fusing the *RUNX1* gene on chromosome 21 with a gene on chromosome 8 called *RUNX1T1* (also known as *ETO*). Because this genetic change affects CBF, the condition is classified as core binding factor AML (CBF-AML).

The resulting fusion protein, RUNX1-ETO, is able to form CBF and attach to DNA, like the normal RUNX1 protein; however, instead of turning genes on, it turns them off. This change in gene activity blocks the maturation (differentiation) of blood cells and leads to the production of abnormal, immature white blood cells called myeloid blasts. While t(8;21) is important for leukemia development, a mutation in one or more additional genes is typically needed for the myeloid blasts to develop into cancerous leukemia cells.

Cytogenetically normal acute myeloid leukemia

Juvenile idiopathic arthritis

Rheumatoid arthritis

Systemic mastocytosis

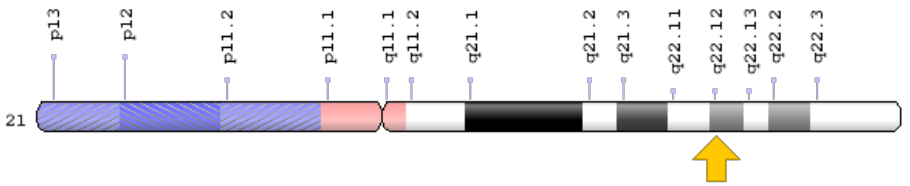
Other disorders

Translocations and other types of mutations involving the *RUNX1* gene have been associated with different types of leukemia and related blood disorders, including acute lymphoblastic leukemia (ALL), chronic myelomonocytic leukemia (CMML), familial platelet disorder with predisposition to acute myeloid leukemia, and myelodysplastic syndromes (MDS). Depending on the type of mutation, these conditions can be related to impaired regulation of gene activity or loss of normal gene function. The *RUNX1* gene mutations associated with these diseases are somatic mutations and are not inherited. They are found only in certain cells of the body.

## Chromosomal Location

Cytogenetic Location: 21q22.12, which is the long (q) arm of chromosome 21 at position 22.12

Molecular Location: base pairs 34,787,801 to 35,049,334 on chromosome 21 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- acute myeloid leukemia 1 protein
- AML1
- AMLCR1
- CBF-alpha-2
- CBFA2
- core-binding factor, runt domain, alpha subunit 2
- oncogene AML-1
- PEA2-alpha B
- PEBP2-alpha B

- PEBP2A2
- PEBP2aB
- polyomavirus enhancer-binding protein 2 alpha B subunit
- runt-related transcription factor 1
- RUNX1_HUMAN
- SL3-3 enhancer factor 1 alpha B subunit
- SL3/AKV core-binding factor alpha B subunit

## Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Structure and Function of Transcriptional Activators
  https://www.ncbi.nlm.nih.gov/books/NBK9904/#_A1003_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RUNX1%5BTI%5D%29+OR+%28runt-related+transcription+factor+1%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+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PLATELET DISORDER, FAMILIAL, WITH ASSOCIATED MYELOID MALIGNANCY
  http://omim.org/entry/601399
- RUNT-RELATED TRANSCRIPTION FACTOR 1
  http://omim.org/entry/151385

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/AML1ID52.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RUNX1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10471
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:861

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/861
- UniProt
  https://www.uniprot.org/uniprot/Q01196

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