RUNX1 gene

RUNX family 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 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

MedlinePlus Genetics provides information about Cytogenetically normal acute myeloid

leukemia

Juvenile idiopathic arthritis

MedlinePlus Genetics provides information about Juvenile idiopathic arthritis

Rheumatoid arthritis

MedlinePlus Genetics provides information about Rheumatoid arthritis

Systemic mastocytosis

MedlinePlus Genetics provides information about 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.

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

Tests Listed in the Genetic Testing Registry

- Tests of RUNX1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=861[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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

- RUNT-RELATED TRANSCRIPTION FACTOR 1; RUNX1 (https://omim.org/entry/151385)
- PLATELET DISORDER, FAMILIAL, WITH ASSOCIATED MYELOID MALIGNANCY; FPDMM (https://omim.org/entry/601399)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/861)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RUNX1[gene])

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Genomic Location

The *RUNX1* gene is found on chromosome 21 (https://medlineplus.gov/genetics/chromosome/21/).

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