

## RUNX1T1 gene

RUNX1 partner transcriptional co-repressor 1

### Normal Function

The *RUNX1T1* gene provides instructions for making a protein commonly referred to as ETO, which helps regulate the activity of genes. ETO is considered a transcriptional corepressor because it turns off (represses) gene activity. It performs this function by attaching (binding) to proteins that normally turn genes on and blocking their activity. It also interacts with other corepressors to help keep genes turned off.

### Health Conditions Related to Genetic Changes

#### Core binding factor acute myeloid leukemia

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

The protein produced from the normal *RUNX1* gene is part of a protein complex known as core binding factor (CBF). As part of CBF, the RUNX1 protein attaches to specific areas of DNA and turns on (activates) genes that are involved in the development of blood cells. Like RUNX1, the fusion protein produced from the t(8;21) translocation, called RUNX1-ETO, attaches to DNA; however, because ETO is involved, the fusion protein turns off genes related to blood cell development instead of turning them on. 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, one or more additional genetic changes are typically needed for the myeloid blasts to develop into cancerous leukemia cells.

### Other Names for This Gene

- acute myelogenous leukemia 1 translocation 1, cyclin-D related
- AML1T1
- CBFA2T1

- CDR
- core-binding factor, runt domain, alpha subunit 2; translocated to, 1; cyclin D-related
- eight twenty one protein
- ETO
- MTG8
- MTG8\_HUMAN
- myeloid translocation gene on 8q22
- protein CBFA2T1
- runt related transcription factor 1; translocated to, 1 (cyclin D related)
- runt-related transcription factor 1; translocated to, 1 (cyclin D-related)
- zinc finger MYND domain-containing protein 2
- ZMYND2

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of RUNX1T1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=862\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=862[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RUNX1T1%5BTI%5D%29+OR+%28ETO%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+1080+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- RUNT-RELATED TRANSCRIPTION FACTOR 1, TRANSLOCATED TO, 1; RUNX1T1 (<https://omim.org/entry/133435>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/862>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RUNX1T1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RUNX1T1[gene]))

## **References**

- Goyama S, Mulloy JC. Molecular pathogenesis of core binding factor leukemia:

current knowledge and future prospects. *Int J Hematol*. 2011 Aug;94(2):126-133.doi: 10.1007/s12185-011-0858-z. Epub 2011 May 3. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21537931>)

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- Lam K, Zhang DE. RUNX1 and RUNX1-ETO: roles in hematopoiesis andleukemogenesis. *Front Biosci (Landmark Ed)*. 2012 Jan 1;17(3):1120-39. doi:10.2741/3977. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22201794>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3433167/>)

## Genomic Location

The *RUNX1T1* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

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