CBFB gene

core-binding factor subunit beta

Normal Function

The *CBFB* gene provides instructions for making a protein called core binding factor beta (CBFβ), which is one piece of a protein complex known as core binding factor (CBF). CBFβ attaches (binds) to one of three related RUNX proteins (RUNX1, RUNX2, or RUNX3) to form different versions of CBF. These protein complexes bind to specific regions of DNA and help turn on (activate) certain genes.

The presence of CBFβ helps the complex bind to DNA and protects the RUNX protein from being broken down. The function of CBF depends on which RUNX protein it includes. Once bound to DNA, the RUNX1 protein controls the activity of genes involved in the development of blood cells (hematopoiesis). The RUNX2 protein regulates genes important for bone cell development and formation of the skeleton. The RUNX3 protein primarily affects genes involved in the development of nerve cells.

Health Conditions Related to Genetic Changes

Core binding factor acute myeloid leukemia

Rearrangements of genetic material affecting the *CBFB* gene are involved in a form of blood cancer known as acute myeloid leukemia (AML). Because the genetic changes affect CBF, the condition is classified as core binding factor AML (CBF-AML). The most common of these rearrangements is an inversion of a region of chromosome 16 (written as inv(16)). An inversion involves breakage of the chromosome in two places; the resulting piece of DNA is reversed and reinserted into the chromosome. Less commonly, a rearrangement known as a translocation occurs between the two copies of chromosome 16 (written as t(16;16)). In this translocation, pieces of DNA from each copy of the chromosome break off and are interchanged. Both types of genetic rearrangement lead to the fusion of parts of two genes on chromosome 16, *CBFB* and *MYH11*. These rearrangements are associated with 5 to 8 percent of cases of AML in adults.

When these rearrangements occur in early blood cells, the function of the RUNX1 protein is particularly affected. The protein produced from the fusion gene, called CBFβ-MYH11, can still bind to RUNX1 to form CBF. However, the function of CBF is impaired. The presence of CBFβ-MYH11 may block binding of CBF to DNA, preventing RUNX1

from controlling gene activity. Alternatively, the MYH11 portion of the fusion protein may interact with other proteins that prevent RUNX1 from controlling gene activity. 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 inv(16) and t(16;16) are 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

- CBF-beta
- core-binding factor beta subunit
- core-binding factor, beta subunit
- PEA2-beta
- PEBB_HUMAN
- PEBP2-beta
- PEBP2B
- polyomavirus enhancer binding protein 2, beta subunit
- polyomavirus enhancer-binding protein 2 beta subunit
- SL3-3 enhancer factor 1 beta subunit
- SL3-3 enhancer factor 1 subunit beta
- SL3/AKV core-binding factor beta subunit

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CBFB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=865[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CBFB%5BTIAB%5D%29+OR+%28core-binding+factor+beta%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CORE-BINDING FACTOR, BETA SUBUNIT; CBFB (https://omim.org/entry/121360)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/865)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CBFB[gene])

References

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Genomic Location

The *CBFB* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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