

BCOR gene

BCL6 corepressor

Normal Function

The *BCOR* gene provides instructions for making a protein known as the BCL6 corepressor. A corepressor is a protein that cannot attach (bind) to DNA by itself, but interacts with other DNA-binding proteins to suppress the activity of certain genes. In this case, the BCL6 corepressor partners with the DNA-binding protein produced from the *BCL6* gene. The *BCL6* gene plays an important role in the function and survival of certain immune system cells.

Researchers have found that the *BCOR* gene is active throughout the body, not just in the immune system. This widespread activity suggests that the BCL6 corepressor has other functions in addition to its interaction with the BCL6 protein. The BCL6 corepressor appears to play a critical role in early embryonic development, including the formation of the eyes and several other tissues and organs. Scientists believe that the BCL6 corepressor may also be involved in specifying the left and right sides of the body in the developing embryo.

Health Conditions Related to Genetic Changes

Oculofaciocardiodental syndrome

Mutations in the *BCOR* gene can cause oculofaciocardiodental (OFCD) syndrome. Some of these mutations delete large amounts of genetic material from the *BCOR* gene, while other mutations alter the gene's instructions such that no BCL6 corepressor protein can be produced. A loss of this protein disrupts the normal development of the eyes and several other organs and tissues before birth.

OFCD syndrome occurs exclusively in females. OFCD syndrome has an X-linked dominant inheritance pattern, which means one altered copy of the *BCOR* gene in each cell is sufficient to cause the condition. The genetic changes that underlie OFCD syndrome prevent the production of any BCL6 corepressor protein. In males, who have only one X chromosome in each cell, these mutations result in a total loss of the BCL6 corepressor. A lack of this protein appears to be lethal very early in development, so no males are born with OFCD syndrome. In females, who have two X chromosomes in each cell, some cells produce a normal amount of BCL6 corepressor protein and other cells produce none. The resulting overall reduction in the amount of this protein leads to

the signs and symptoms of OFCD syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Microphthalmia

MedlinePlus Genetics provides information about Microphthalmia

Other Names for This Gene

- 5830466J11Rik
- 8430401K06Rik
- ANOP2
- BCL-6 interacting corepressor
- BCL6 co-repressor
- BCOR_HUMAN
- FLJ20285
- FLJ38041
- KIAA1575
- MAA2
- MCOPS2
- MGC131961
- MGC71031

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of BCOR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54880\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54880[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28BCOR%5BTIAB%5D%29+OR+%28BCL6+co-repressor%5BTIAB%5D%29%29+OR+%28MAA2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22Iast+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- BCL6 COREPRESSOR; BCOR (<https://omim.org/entry/300485>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/54880>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=BCOR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=BCOR[gene]))

References

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Genomic Location

The *BCOR* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

Last updated May 1, 2008