



## 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

#### Lenz microphthalmia syndrome

A mutation in the *BCOR* gene has been found in one family with Lenz microphthalmia syndrome. This mutation changes a single protein building block (amino acid) in the BCL6 corepressor, which alters the protein's structure. Specifically, the mutation replaces the amino acid proline with the amino acid leucine at position 85 (written as Pro85Leu). Because the BCL6 corepressor plays an essential role in early development, a defective version of the protein disrupts the normal formation of the eyes and several other organs and tissues before birth.

The *BCOR* gene is on the X chromosome (one of the two sex chromosomes) and the inheritance of Lenz microphthalmia is described as X-linked recessive. X-linked recessive disorders occur much more commonly in males than in females. Lenz microphthalmia syndrome has been found only in males.

#### Oculofaciocardiodental syndrome

Mutations in the *BCOR* gene can also 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.

Unlike Lenz microphthalmia syndrome, which has been found only in males, 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.

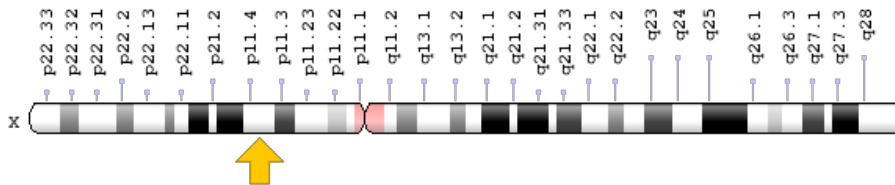
Coloboma

Microphthalmia

### Chromosomal Location

Cytogenetic Location: Xp11.4, which is the short (p) arm of the X chromosome at position 11.4

Molecular Location: base pairs 40,051,246 to 40,177,390 on the X chromosome (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

### 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**

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Eucaryotic gene regulatory proteins often assemble into complexes on DNA  
<https://www.ncbi.nlm.nih.gov/books/NBK26872/#A1300>

### Clinical Information from GeneReviews

- Lenz Microphthalmia Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1521>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?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+%22last+3600+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- BCL6 COREPRESSOR  
<http://omim.org/entry/300485>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_BCOR.html](http://atlasgeneticsoncology.org/Genes/GC_BCOR.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=BCOR%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:20893](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:20893)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:54880>

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/54880>
- UniProt  
<https://www.uniprot.org/uniprot/Q6W2J9>

### Sources for This Summary

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20301694>

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<https://ghr.nlm.nih.gov/gene/BCOR>

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