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 it 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 corepressor appears to play a critical role in early 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. Research also shows that this protein may regulate development by influencing the self-destruction of cells that are damaged or no longer needed (apoptosis).

Health Conditions Related to Genetic Changes

Oculofaciocardiodental syndrome

Variants (also called mutations) in the *BCOR* gene can cause oculofaciocardiodental (OFCD) syndrome. This condition affects the development of the eyes (oculo-), facial features (facio-), heart (cardio-), and teeth (dental). Some of these variants delete large amounts of genetic material from the *BCOR* gene, while other variants 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 (who have two X chromosomes). OFCD syndrome has an X-linked dominant inheritance pattern, which means that 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. As a result, affected individuals have about half of the normal amount of this protein, which leads to the signs and symptoms of OFCD syndrome.

In males (who have only one X chromosome), these variants would prevent the production of any BCL6 corepressor protein. A complete lack of this protein is thought to

be lethal very early in development.

Anophthalmia/Microphthalmia

MedlinePlus Genetics provides information about Anophthalmia/Microphthalmia

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- BCL-6 interacting corepressor
- BCL6 co-repressor
- BCOR_HUMAN

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])

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+%22last+3600+days%22%5Bdp%5D)

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])

References

- Hilton EN, Manson FD, Urquhart JE, Johnston JJ, Slavotinek AM, Hedera P,Stattin EL, Nordgren A, Biesecker LG, Black GC. Left-sided embryonic expressionof the

BCL-6 corepressor, BCOR, is required for vertebrate lateralitydetermination. Hum Mol Genet. 2007 Jul 15;16(14):1773-82. doi:10.1093/hmg/ddm125. Epub 2007 May 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17517692)

- Horn D, Chyrek M, Kleier S, Luttgen S, Bolz H, Hinkel GK, Korenke GC, Riess A, Schell-Apacik C, Tinschert S, Wieczorek D, Gillessen-Kaesbach G, Kutsche K. Novelmutations in BCOR in three patients with oculo-facio-cardio-dental syndrome, butnone in Lenz microphthalmia syndrome. Eur J Hum Genet. 2005 May;13(5):563-9. doi:10.1038/sj.ejhg.5201391. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15770227)
- Huynh KD, Fischle W, Verdin E, Bardwell VJ. BCoR, a novel corepressor involvedin BCL-6 repression. Genes Dev. 2000 Jul 15;14(14):1810-23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10898795) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC316791/)
- Ng D, Thakker N, Corcoran CM, Donnai D, Perveen R, Schneider A, Hadley DW, Tifft C, Zhang L, Wilkie AO, van der Smagt JJ, Gorlin RJ, Burgess SM, BardwellVJ, Black GC, Biesecker LG. Oculofaciocardiodental and Lenz microphthalmiasyndromes result from distinct classes of mutations in BCOR. Nat Genet. 2004Apr;36(4):411-6. doi: 10.1038/ng1321. Epub 2004 Mar 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15004558)
- Ragge N, Isidor B, Bitoun P, Odent S, Giurgea I, Cogne B, Deb W, Vincent M, LeGall J, Morton J, Lim D; DDD Study; Le Meur G, Zazo Seco C, Zafeiropoulou D, BaxD, Zwijnenburg P, Arteche A, Swafiri ST, Cleaver R, McEntagart M, Kini U, NewmanW, Ayuso C, Corton M, Herenger Y, Jeanne M, Calvas P, Chassaing N. Expanding thephenotype of the X-linked BCOR microphthalmia syndromes. Hum Genet. 2019Sep;138(8-9):1051-1069. doi: 10.1007/s00439-018-1896-x. Epub 2018 Jul 4. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/29974297)

Genomic Location

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

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