

ALX1 gene

ALX homeobox 1

Normal Function

The *ALX1* gene provides instructions for making a protein that is a member of the homeobox protein family. Homeobox proteins direct the formation of body structures during early embryonic development. The ALX1 protein is necessary for normal development of the head and face, particularly the formation of the eyes, nose, and mouth, which begins around the fourth week of development. The ALX1 protein is a transcription factor, which means that it attaches (binds) to DNA and controls the activity of certain genes. Specifically, the protein controls the activity of genes that regulate cell growth and division (proliferation) and movement (migration), ensuring that cells grow and stop growing at specific times and that they are positioned correctly during development.

Health Conditions Related to Genetic Changes

Frontonasal dysplasia

At least three mutations in the *ALX1* gene have been found to cause frontonasal dysplasia. *ALX1* gene mutations cause a form of the disorder called frontonasal dysplasia type 3, which is characterized by severe malformations of the structures at the center of the face. *ALX1* gene mutations that cause this condition alter the protein's structure and impair its ability to bind to DNA and regulate gene function. As a result, the proliferation and migration of cells during development is not controlled, which can lead to small or missing eyes, openings (clefts) in the nose or mouth, and other severe facial malformations characteristic of frontonasal dysplasia type 3.

Other Names for This Gene

- ALX homeobox protein 1
- CART-1
- CART1
- cartilage paired-class homeoprotein 1
- epididymis luminal protein 23
- FND3

- HEL23

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALX1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8092\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8092[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ALX1%5BTIAB%5D%29+OR+%28%28CART1%5BTIAB%5D%29+OR+%28CART-1%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+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ARISTALESS-LIKE HOMEBOX 1; ALX1 (<https://omim.org/entry/601527>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8092>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ALX1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ALX1[gene]))

References

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- McGonnell IM, Graham A, Richardson J, Fish JL, Depew MJ, Dee CT, Holland PW, Takahashi T. Evolution of the Alx homeobox gene family: parallel retention and independent loss of the vertebrate Alx3 gene. *Evol Dev.* 2011 Jul-Aug;13(4):343-51. doi: 10.1111/j.1525-142X.2011.00489.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21740507>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3166657/>)
- Uz E, Alanay Y, Aktas D, Vargel I, Gucer S, Tuncbilek G, von Eggeling F, Yilmaz E, Deren O, Posorski N, Ozdag H, Liehr T, Balci S, Alikasifoglu M, Wollnik B, Akarsu NA. Disruption of ALX1 causes extreme microphthalmia and severe facial clefting: expanding the spectrum of autosomal-recessive ALX-related frontonasal dysplasia. *Am J Hum Genet.* 2010 May 14;86(5):789-96. doi:10.1016/j.ajhg.2010.04.002. Epub 2010 May 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20451171>) or

Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2869009/>)

Genomic Location

The *ALX1* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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