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

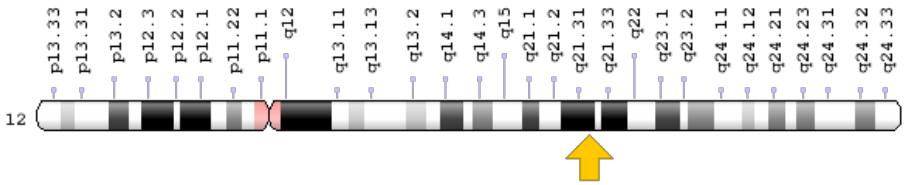
Frontonasal dysplasia

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.

Chromosomal Location

Cytogenetic Location: 12q21.31, which is the long (q) arm of chromosome 12 at position 21.31

Molecular Location: base pairs 85,280,220 to 85,301,784 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Jasper's Basic Mechanisms of the Epilepsies (fourth edition, 2012): Molecular Biology of ARX
  https://www.ncbi.nlm.nih.gov/books/NBK98176/#marsh.s3

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

Catalog of Genes and Diseases from OMIM

- ARISTALESS-LIKE HOMEOBOX 1
  http://omim.org/entry/601527

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALX1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALX1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:1494
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8092
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8092
- UniProt
  https://www.uniprot.org/uniprot/Q15699

Sources for This Summary

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