



ARSL gene

arylsulfatase E

Normal Function

The *ARSL* gene provides instructions for making an enzyme called arylsulfatase E. This enzyme is part of a group known as sulfatases, which are enzymes that help process molecules that contain chemical groups known as sulfates. Sulfatases play important roles in cartilage and bone development.

Within cells, arylsulfatase E is located in the Golgi apparatus, a structure that modifies newly produced enzymes and other proteins. The function of this enzyme is unknown, although researchers believe it participates in a chemical pathway involving vitamin K. Evidence suggests that vitamin K normally plays a role in bone growth and maintenance of bone density.

Health Conditions Related to Genetic Changes

X-linked chondrodysplasia punctata 1

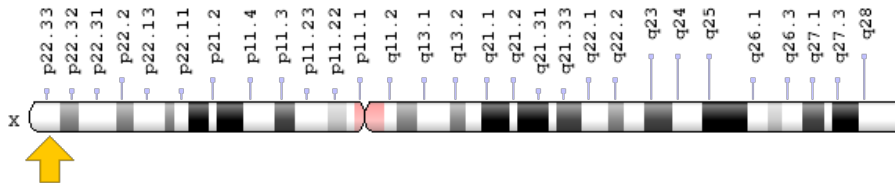
Genetic changes involving the *ARSL* gene are responsible for X-linked chondrodysplasia punctata 1, a disorder of bone and cartilage development that occurs almost exclusively in males. Between 60 and 75 percent of males with the characteristic features of this condition have a mutation within the *ARSL* gene. At least 18 mutations have been found in affected individuals; these genetic changes reduce or eliminate the function of arylsulfatase E. Another 25 percent of affected males have a small deletion of genetic material from the region of the X chromosome that contains the *ARSL* gene. These individuals are missing the entire gene, so their cells produce no functional arylsulfatase E.

It is unclear how a shortage of arylsulfatase E disrupts the development of bones and cartilage and leads to the characteristic features of X-linked chondrodysplasia punctata 1.

Chromosomal Location

Cytogenetic Location: Xp22.33, which is the short (p) arm of the X chromosome at position 22.33

Molecular Location: base pairs 2,934,521 to 2,968,312 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARSE
- ARSE_HUMAN
- arylsulfatase E (chondrodysplasia punctata 1)
- CDPX
- CDPX1
- CDPXR
- MGC163310

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Osteogenesis: The Development of Bones
<https://www.ncbi.nlm.nih.gov/books/NBK10056/>

Clinical Information from GeneReviews

- Chondrodysplasia Punctata 1, X-Linked
<https://www.ncbi.nlm.nih.gov/books/NBK1544>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARSE%5BTIAB%5D%29+OR+%28arylsulfatase+E%5BTIAB%5D%29%29+OR+%28CDPX1%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

- ARYLSULFATASE E
<http://omim.org/entry/300180>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ARSE.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ARSL%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:719
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:415>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/415>
- UniProt
<https://www.uniprot.org/uniprot/P51690>

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