

ARSL gene

arylsulfatase L

Normal Function

The *ARSL* gene provides instructions for making an enzyme called arylsulfatase L. 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 L 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 L. 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 L.

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

Other Names for This Gene

- ARSE
- ARSE_HUMAN
- arylsulfatase E

- CDPX
- CDPX1
- CDPXR
- MGC163310

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ARSL ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=415\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=415[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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 L; ARSL (<https://omim.org/entry/300180>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/415>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ARSL\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ARSL[gene]))

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Genomic Location

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

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