



ARSB gene

arylsulfatase B

Normal Function

The *ARSB* gene provides instructions for producing an enzyme called arylsulfatase B, which is involved in the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Specifically, arylsulfatase B removes a chemical group known as a sulfate from two GAGs called dermatan sulfate and chondroitin sulfate. Arylsulfatase B is located in lysosomes, compartments within cells that digest and recycle different types of molecules.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type VI

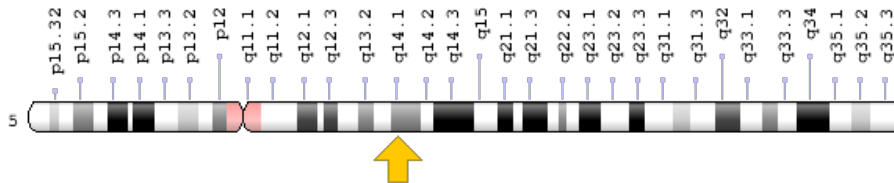
More than 130 mutations in the *ARSB* gene have been found to cause mucopolysaccharidosis type VI (MPS VI). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS VI reduce or eliminate the function of arylsulfatase B. It usually cannot be determined whether a certain mutation will cause severe or mild MPS VI; however, mutations known to result in the complete absence of arylsulfatase B activity cause severe signs and symptoms.

The lack of arylsulfatase B activity leads to the accumulation of GAGs within lysosomes. Conditions such as MPS VI that cause molecules to build up inside the lysosomes are called lysosomal storage disorders. The accumulation of GAGs within lysosomes increases the size of cells, which is why many tissues and organs are enlarged in this disorder. Researchers believe that the buildup of GAGs may also interfere with the functions of other proteins inside lysosomes, triggering inflammation and cell death.

Chromosomal Location

Cytogenetic Location: 5q14.1, which is the long (q) arm of chromosome 5 at position 14.1

Molecular Location: base pairs 78,777,209 to 78,986,087 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARSB_HUMAN
- arylsulfatase B isoform 1 precursor
- ASB
- chondroitinase
- chondroitinsulfatase
- G4S
- MPS6
- N-acetylgalactosamine-4-sulfatase
- N-acetylgalactosamine 4-sulfate sulfohydrolase

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARSB%5BTIAB%5D%29+OR+%28arylsulfatase+B%5BTIAB%5D%29%29+OR+%28N-acetylgalactosamine-4-sulfatase%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 B
<http://omim.org/entry/611542>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ARSB.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ARSB%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:714
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:411>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/411>
- UniProt
<https://www.uniprot.org/uniprot/P15848>

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