ARSB gene
aryl sulfatase B

Normal Function

The ARSB gene provides instructions for producing an enzyme called aryl sulfatase B, which is involved in the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Specifically, aryl sulfatase 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 aryl sulfatase 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 aryl sulfatase B activity cause severe signs and symptoms.

The lack of aryl sulfatase 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+OR+%28N-acetylgalactosamine-4-sulfatase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

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 Family: Sulfatases
  https://www.genenames.org/cgi-bin/genefamilies/set/410

- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=714

- NCBI Gene

- UniProt
  http://www.uniprot.org/uniprot/P15848

Sources for This Summary

- OMIM: ARYLSULFATASE B
  http://omim.org/entry/611542


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18406185

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17458871

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11668612

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19531206
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