ARSA gene
arylsulfatase A

Normal Function

The ARSA gene provides instructions for making the enzyme arylsulfatase A. This enzyme is located in cellular structures called lysosomes, which are the cell's recycling centers. Within lysosomes, arylsulfatase A helps process substances known as sulfatides. Sulfatides are a subgroup of sphingolipids, a category of fats that are important components of cell membranes. Sulfatides are abundant in the nervous system's white matter, consisting of nerve fibers covered by myelin. Myelin, made up of multiple layers of membranes, insulates and protects nerves.

Health Conditions Related to Genetic Changes

Metachromatic leukodystrophy

More than 110 mutations that cause metachromatic leukodystrophy, a disorder that causes deterioration of nervous system functions, have been identified in the ARSA gene. These mutations greatly reduce the activity of arylsulfatase A. Severe disruption in arylsulfatase A activity interferes with the breakdown of sulfatides. As a result, these substances can accumulate to toxic levels in the nervous system. The buildup of sulfatides gradually destroys the cells that produce myelin, the covering that protects nerves and promotes the efficient transmission of nerve impulses. Destruction of myelin leads to the loss of white matter (leukodystrophy) and impairment of nervous system function, resulting in the signs and symptoms of metachromatic leukodystrophy.

In some cases, individuals with very low arylsulfatase A activity show no signs or symptoms of metachromatic leukodystrophy. This condition, called pseudoarylsulfatase deficiency, seems to be caused by specific variations of the ARSA gene. These variations are present in as many as 5 to 10 percent of Europeans and North Americans.
Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 50,622,754 to 50,628,152 on chromosome 22 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARSA_HUMAN
- cerebroside 3-sulfatase
- Cerebrosidase-3-sulfate 3-sulfohydrolase
- Cerebroside-Sulfatase
- MLD
- sulfatidase

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Lysosomal Disease
  https://www.ncbi.nlm.nih.gov/books/NBK28215/
  https://www.ncbi.nlm.nih.gov/books/NBK20729/#A1383

Clinical Information from GeneReviews

- Arylsulfatase A Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1130
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARSA%5BTIAB%5D%29+OR+%28arylsulfatase+A%5BTIAB%5D%29+OR+%28arylsulphatase+A%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ARYLSULFATASE A
  http://omim.org/entry/607574

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ARSA.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:410
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P15289

Sources for This Summary

- OMIM: ARYLSULFATASE A
  http://omim.org/entry/607574
- Basic Neurochemistry (sixth edition, 1999): Lysosomal Disease
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16966551

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21167507

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