

## 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.

### Other Names for This Gene

- ARSA\_HUMAN
- cerebroside 3-sulfatase
- Cerebroside-3-sulfate 3-sulfohydrolase

- Cerebroside-Sulfatase
- MLD
- sulfatidase

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ARSA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=410\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=410[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

### Catalog of Genes and Diseases from OMIM

- ARYLSULFATASE A; ARSA (<https://omim.org/entry/607574>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/410>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ARSA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ARSA[gene]))

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## Genomic Location

The ARSA gene is found on chromosome 22 (<https://medlineplus.gov/genetics/chromosome/22/>).

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