ASL gene

argininosuccinate lyase

Normal Function

The *ASL* gene provides instructions for making the enzyme argininosuccinate lyase. This enzyme primarily participates in the urea cycle, a series of reactions that occur in liver cells. The urea cycle processes excess nitrogen, generated when protein is used by the body, to make a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia.

The specific role of the argininosuccinate lyase enzyme is to start the reaction in which the amino acid arginine, a building block of proteins, is produced from argininosuccinate, the molecule that carries the waste nitrogen collected earlier in the urea cycle. The arginine is later broken down into urea, which is excreted, and ornithine, which restarts the urea cycle.

In cells throughout the body, the argininosuccinate lyase enzyme is also involved in moving (transporting) arginine into cells to make a compound called nitric oxide. Nitric oxide is important for regulating blood flow and blood pressure.

Health Conditions Related to Genetic Changes

Argininosuccinic aciduria

More than 130 mutations in the *ASL* gene have been found to cause argininosuccinic aciduria. In some cases, a short sequence of DNA is deleted from the gene. Other mutations replace one protein building block (amino acid) with another amino acid in the argininosuccinate lyase enzyme. In people of Arab ancestry, two common mutations replace the amino acid glutamine with a premature stop signal at position 116 (written as Gln116Ter or Q116*) or position 354 (written as Gln354Ter or Q354*) in the argininosuccinate lyase enzyme. Mutations in the *ASL* gene may result in an argininosuccinate lyase enzyme that is unstable, misshapen, or quickly broken down.

If the argininosuccinate lyase enzyme is misshapen or missing, it cannot fulfill its role in the urea cycle. Arginine is not produced, excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. This buildup of ammonia damages the brain and other tissues and causes neurological problems and other signs and symptoms of argininosuccinic aciduria. It is unclear how a lack of arginine contributes to

the features of this condition.

Other Names for This Gene

- Argininosuccinase
- Arginosuccinase
- arginosuccinate lyase
- ARLY_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ASL (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=435[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ASL%5BTIAB%5D%29+OR+%28argininosuccinate+lyase%5BTIAB%5D%29%29+OR+%28%28Argininosuccinase%5BTIAB%5D%29+OR+%28Arginosuccinase%5BTIAB%5D%29+OR+%28arginosuccinate+lyase%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ARGININOSUCCINATE LYASE; ASL (https://omim.org/entry/608310)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/435)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ASL[gene])

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Genomic Location

The *ASL* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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