ASL gene
argininosuccinate lyase

Normal Function
The ASL gene provides instructions for making the protein argininosuccinate lyase. This enzyme 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 ASL 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 Restart restarts the urea cycle.

Health Conditions Related to Genetic Changes
Argininosuccinic aciduria

More than 30 different mutations in the ASL gene have been identified worldwide. 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 descent, two common mutations replace the amino acid glutamine with a premature stop signal at position 116 (written as Gln116Ter or Q116X) or position 354 (written as Gln354Ter or Q354X) in the argininosuccinate lyase enzyme. Mutations in the ASL gene may result in an argininosuccinate lyase enzyme that is unstable or the wrong shape.

The shape of an enzyme affects its ability to control a chemical reaction. If the argininosuccinate lyase enzyme is misshapen or missing, it cannot fulfill its role in the urea cycle. Excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. Ammonia is toxic, especially to the nervous system, so this accumulation causes neurological problems and other signs and symptoms of argininosuccinic aciduria.
Chromosomal Location

Cytogenetic Location: 7q11.21, which is the long (q) arm of chromosome 7 at position 11.21

Molecular Location: base pairs 66,075,819 to 66,093,576 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• Argininosuccinase
• Arginosuccinase
• arginosuccinate lyase
• ARLY_HUMAN

Additional Information & Resources

Educational Resources

• Biochemistry (fifth edition, 2002): Ammonium Ion is Converted into Urea in Most Terrestrial Vertebrates.
  https://www.ncbi.nlm.nih.gov/books/NBK22450/

Clinical Information from GeneReviews

• Argininosuccinate Lyase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK51784

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ASL%5BTIAB%5D%29+OR+%28argininosuccinate+lyase%5BTIAB%5D%29+OR%28argininosuccinate+lyase%5BTIAB%5D%29%29+AND+%28Genes%5BMH%29+OR+%28Genetic+Phenomena%5BMH%29+AND+english%5Bl+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D
ARGININOSUCCINATE LYASE
http://omim.org/entry/608310

Research Resources

Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ASL.html

ClinVar
https://www.ncbi.nlm.nih.gov/clinvar?term=ASL%5Bgene%5D

HGNC Gene Symbol Report

Monarch Initiative
https://monarchinitiative.org/gene/NCBIGene:435

NCBI Gene

UniProt
https://www.uniprot.org/uniprot/P04424

Sources for This Summary

OMIM: ARGININOSUCCINATE LYASE
http://omim.org/entry/608310

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16435180

Biochemistry (fifth edition, 2002): Ammonium Ion is Converted into Urea in Most Terrestrial Vertebrates.
https://www.ncbi.nlm.nih.gov/books/NBK22450/

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16941645

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12384776

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12559843

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC23030/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11092456

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11747433

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