ARG1 gene
arginase 1

Normal Function

The ARG1 gene provides instructions for producing the enzyme arginase. This enzyme participates in the urea cycle, a series of reactions that occurs in liver cells. The urea cycle processes excess nitrogen, which is generated when proteins and their building blocks (amino acids) are used by the body. Through the urea cycle, excess nitrogen is made into a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic.

Arginase controls the last step of the urea cycle, a reaction in which nitrogen is removed from the amino acid arginine and processed into urea for excretion from the body. A compound called ornithine is also produced in this reaction; it is needed for the urea cycle to repeat.

Health Conditions Related to Genetic Changes

Arginase deficiency

Approximately 12 mutations have been identified in the ARG1 gene. A mutated ARG1 gene may result in an arginase enzyme that is unstable, shorter than usual or the wrong shape, or may prevent the enzyme from being produced at all.

The shape of an enzyme affects its ability to control a chemical reaction. If the arginase 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 and arginine accumulate in the body. Ammonia is toxic, especially to the nervous system, and the accumulation of ammonia and arginine are believed to cause the neurological problems and other signs and symptoms of arginase deficiency.
Chromosomal Location

Cytogenetic Location: 6q23.2, which is the long (q) arm of chromosome 6 at position 23.2

Molecular Location: base pairs 131,573,159 to 131,584,332 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• A-I
• ARG1_HUMAN
• arginase, liver
• arginase, type I

Additional Information & Resources

Educational Resources

• Biochemistry (5th edition, 2002): Ammonium Ion is Converted into Urea in Most Terrestrial Vertebrates.
  https://www.ncbi.nlm.nih.gov/books/NBK22450/

Clinical Information from GeneReviews

• Arginase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1159

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARG1%5BTIAB%5D %29+OR+%28arginase,+liver%5BTIAB%5D%29+OR+%28arginase,+type+I%5BTIAB%5D%29+OR+%28A-I%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 +360+days%22%5Bdp%5D

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http://omim.org/entry/608313

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ARG1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ARG1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=663
- Monarch Initiative
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P05089

Sources for This Summary

- OMIM: ARGINASE 1
  http://omim.org/entry/608313
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9686347
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12052859
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC133904/
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