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. The urea cycle produces a compound called urea from excess nitrogen. Urea is then excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic.

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

Health Conditions Related to Genetic Changes

Arginase deficiency

Many variants (also called mutations) have been identified in the *ARG1* gene. Arginase deficiency causes ammonia to accumulate gradually in the blood. The nervous system is especially sensitive to the effects of excess ammonia. Changes in the *ARG1* gene can result in an arginase enzyme that is unstable, shorter than usual, or the wrong shape. Variants in the *ARG1* gene may also 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. High levels of ammonia and arginine are believed to cause the neurological problems and other signs and symptoms of arginase deficiency.

Other Names for This Gene

- A-I
- ARGI1_HUMAN
- arginase, liver
- arginase, type I

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ARG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=383[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ARG1%5BTIAB%5D%29+OR+%28arginase,+liver%5BTIAB%5D%29%29+OR+%28%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)

Catalog of Genes and Diseases from OMIM

- ARGINASE 1; ARG1 (https://omim.org/entry/608313)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/383)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ARG1[gene])

References

- Iyer R, Jenkinson CP, Vockley JG, Kern RM, Grody WW, Cederbaum S. The humanarginases and arginase deficiency. J Inherit Metab Dis. 1998;21 Suppl 1:86-100.doi: 10.1023/a:1005313809037. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9686347)
- Iyer RK, Yoo PK, Kern RM, Rozengurt N, Tsoa R, O'Brien WE, Yu H, Grody WW,Cederbaum SD. Mouse model for human arginase deficiency. Mol Cell Biol. 2002Jul;22(13):4491-8. doi: 10.1128/MCB.22.13.4491-4498.2002. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12052859) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC133904/)
- Vockley JG, Goodman BK, Tabor DE, Kern RM, Jenkinson CP, Grody WW, CederbaumSD. Loss of function mutations in conserved regions of the human arginase I gene.Biochem Mol Med. 1996 Oct;59(1):44-51. doi: 10.1006/bmme.1996.0063. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8902193)

Genomic Location

The *ARG1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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