APRT gene

adenine phosphoribosyltransferase

Normal Function

The *APRT* gene provides instructions for making an enzyme called adenine phosphoribosyltransferase (APRT). This enzyme is produced in all cells and is part of the purine salvage pathway, which recycles a group of DNA building blocks (nucleotides) called purines to make other molecules. The APRT enzyme helps to recycle the purine adenine to make a molecule called adenosine monophosphate (AMP). This conversion occurs when AMP is needed as a source of energy for cells.

Health Conditions Related to Genetic Changes

Adenine phosphoribosyltransferase deficiency

At least 40 mutations in the *APRT* gene have been found to cause adenine phosphoribosyltransferase (APRT) deficiency, a condition that affects the kidneys and urinary tract. Most of these mutations change single protein building blocks (amino acids) in the APRT enzyme. The mutations that cause APRT deficiency are categorized into two groups known as the *APRT**J allele and the *APRT**Q0 allele. The *APRT**J allele consists of one mutation that replaces the amino acid methionine with the amino acid threonine at position 136 in the APRT enzyme (written as Met136Thr or M136T). This mutation reduces the function of the enzyme. The M136T mutation occurs almost exclusively in Japanese individuals with the condition; most affected individuals have this mutation on both copies of the APRT gene in each cell. The *APRT**Q0 allele consists of all other *APRT* gene mutations. The most common of these mutations (written IVS4+2insT) alters the genetic instructions used to make the enzyme, resulting in an abnormally short, nonfunctional enzyme. This mutation is estimated to occur in 40 percent of affected Europeans.

APRT gene mutations lead to a lack of functional enzyme that prevents the conversion of adenine to AMP. As a result, adenine is converted to another molecule called 2,8-dihydroxyadenine (2,8-DHA). 2,8-DHA crystallizes in urine, forming stones in the kidneys and urinary tract. As a result, kidney function can decline, which may lead to end-stage renal disease (ESRD), a life-threatening failure of kidney function.

Other Names for This Gene

- AMP diphosphorylase
- AMP pyrophosphorylase
- APRTase
- APT_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of APRT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=353[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28APRT%5BTIAB%5D%29+OR+%28adenine+phosphoribosyltransferase%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ADENINE PHOSPHORIBOSYLTRANSFERASE; APRT (https://omim.org/entry/102600)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/353)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=APRT[gene])

References

- Bollee G, Dollinger C, Boutaud L, Guillemot D, Bensman A, Harambat J, DeteixP, Daudon M, Knebelmann B, Ceballos-Picot I. Phenotype and genotypecharacterization of adenine phosphoribosyltransferase deficiency. J Am SocNephrol. 2010 Apr;21(4):679-88. doi: 10.1681/ASN.2009080808. Epub 2010 Feb 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20150536) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2844298/)
- Bollee G, Harambat J, Bensman A, Knebelmann B, Daudon M, Ceballos-Picot I. Adenine phosphoribosyltransferase deficiency. Clin J Am Soc Nephrol. 2012Sep;7(9):1521-7. doi: 10.2215/CJN.02320312. Epub 2012 Jun 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22700886)

- Silva M, Silva CH, Iulek J, Thiemann OH. Three-dimensional structure of humanadenine phosphoribosyltransferase and its relation to DHA-urolithiasis. Biochemistry. 2004 Jun 22;43(24):7663-71. doi: 10.1021/bi0360758. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15196008)

Genomic Location

The *APRT* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

Last updated October 1, 2012