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.
Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 88,809,339 to 88,811,928 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AMP diphosphorylase
- AMP pyrophosphorylase
- APRTase
- APT_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Purine Release and Metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK28118/

- Biochemistry (fifth edition, 2002): Purine Bases Can Be Synthesized de Novo or Recycled by Salvage Pathways
  https://www.ncbi.nlm.nih.gov/books/NBK22385/

Clinical Information from GeneReviews

- Adenine Phosphoribosyltransferase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK100238

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/102600

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_APRT.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=APRT%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:353
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P07741

Sources for This Summary

- OMIM: ADENINE PHOSPHORIBOSYLTRANSFERASE
  http://omim.org/entry/102600
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2844298/

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