



Adenine phosphoribosyltransferase deficiency

Adenine phosphoribosyltransferase (APRT) deficiency is an inherited condition that affects the kidneys and urinary tract. The most common feature of this condition is recurrent kidney stones; urinary tract stones are also a frequent symptom. Kidney and urinary tract stones can create blockages in the urinary tract, causing pain during urination and difficulty releasing urine.

Affected individuals can develop features of this condition anytime from infancy to late adulthood. When the condition appears in infancy, the first sign is usually the presence of tiny grains of reddish-brown material in the baby's diaper caused by the passing of stones. Later, recurrent kidney and urinary tract stones can lead to problems with kidney function beginning as early as mid- to late childhood. Approximately half of individuals with APRT deficiency first experience signs and symptoms of the condition in adulthood. The first features in affected adults are usually kidney stones and related urinary problems. Other signs and symptoms of APRT deficiency caused by kidney and urinary tract stones include fever, urinary tract infection, blood in the urine (hematuria), abdominal cramps, nausea, and vomiting.

Without treatment, kidney function can decline, which may lead to end-stage renal disease (ESRD). ESRD is a life-threatening failure of kidney function that occurs when the kidneys are no longer able to filter fluids and waste products from the body effectively.

The features of this condition and their severity vary greatly among affected individuals, even among members of the same family. It is estimated that 15 to 20 percent of people with APRT deficiency do not have any signs or symptoms of the condition.

Frequency

APRT deficiency is estimated to affect 1 in 27,000 people in Japan. The condition is rarer in Europe, where it is thought to affect 1 in 50,000 to 100,000 people. The prevalence of APRT deficiency outside these populations is unknown.

Causes

Mutations in the *APRT* gene cause APRT deficiency. This gene provides instructions for making APRT, an enzyme that helps to convert a DNA building block (nucleotide) called adenine to a molecule called adenosine monophosphate (AMP). This conversion occurs when AMP is needed as a source of energy for cells.

APRT gene mutations lead to the production of an abnormal APRT enzyme with reduced function or prevent the production of any enzyme. A lack of functional enzyme impairs 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. 2,8-DHA crystals are brownish in color, which explains why affected infants frequently have dark urine stains in their diapers. 2,8-DHA is toxic to kidneys, which may explain the possible decline in kidney function and the progression to ESRD.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- 2,8-dihydroxyadenine urolithiasis
- 2,8-dihydroxyadeninuria
- APRT deficiency
- DHA crystalline nephropathy

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: Adenine phosphoribosyltransferase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268120/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22adenine+phosphoribosyltransferase+deficiency%22>

Other Diagnosis and Management Resources

- Boston Children's Hospital: Kidney Stones
<http://www.childrenshospital.org/conditions-and-treatments/conditions/k/kidney-stones>
- GeneReview: Adenine Phosphoribosyltransferase Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK100238>

Additional Information & Resources

Genetic and Rare Diseases Information Center

- Adenine phosphoribosyltransferase deficiency
<https://rarediseases.info.nih.gov/diseases/546/adenine-phosphoribosyltransferase-deficiency>

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Kidney Failure
<https://www.niddk.nih.gov/health-information/kidney-disease/kidney-failure>
- National Institute of Diabetes and Digestive and Kidney Diseases: Kidney Stones
<https://www.niddk.nih.gov/health-information/urologic-diseases/kidney-stones>
- National Institute of Diabetes and Digestive and Kidney Diseases: Your Kidneys and How They Work
<https://www.niddk.nih.gov/health-information/kidney-disease/kidneys-how-they-work>

Educational Resources

- Boston Children's Hospital: Kidney Failure in Children
<http://www.childrenshospital.org/conditions-and-treatments/conditions/k/kidney-failure>
- Johns Hopkins Medicine: Kidney Stones
<https://www.hopkinsmedicine.org/health/conditions-and-diseases/kidney-stones>
- MalaCards: adenine phosphoribosyltransferase deficiency
https://www.malacards.org/card/adenine_phosphoribosyltransferase_deficiency
- Merck Manual for Patients and Caregivers: Stones in the Urinary Tract
<https://www.merckmanuals.com/home/kidney-and-urinary-tract-disorders/stones-in-the-urinary-tract/stones-in-the-urinary-tract>
- Orphanet: Adenine phosphoribosyltransferase deficiency
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=976

Patient Support and Advocacy Resources

- American Kidney Fund
<http://www.kidneyfund.org/kidney-disease/>
- Metabolic Support (UK)
<https://www.metabolicsupportuk.org/>
- National Kidney Foundation
<https://www.kidney.org/>
- Rare Kidney Stone Consortium
<http://www.rarekidneystones.org/dha/>

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=%28adenine+phosphoribosyl+transferase+deficiency%5BTIAB%5D%29+OR+%28aprt+deficiency%5BTIAB%5D%29+OR+%282,8-dihydroxyadeninuria%5BTIAB%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 DEFICIENCY
<http://omim.org/entry/614723>

Sources for This Summary

- Bollée G, Dollinger C, Boutaud L, Guillemot D, Bensman A, Harambat J, Deteix P, Daudon M, Knebelmann B, Ceballos-Picot I. Phenotype and genotype characterization of adenine phosphoribosyltransferase deficiency. *J Am Soc Nephrol.* 2010 Apr;21(4):679-88. doi: 10.1681/ASN.2009080808. Epub 2010 Feb 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20150536>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2844298/>
- Bollée G, Harambat J, Bensman A, Knebelmann B, Daudon M, Ceballos-Picot I. Adenine phosphoribosyltransferase deficiency. *Clin J Am Soc Nephrol.* 2012 Sep;7(9):1521-7. Epub 2012 Jun 14. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22700886>
- Edvardsson VO, Palsson R, Sahota A. Adenine Phosphoribosyltransferase Deficiency. 2012 Aug 30 [updated 2015 Jun 18]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK100238/>
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22934314>
- Harambat J, Bollée G, Daudon M, Ceballos-Picot I, Bensman A; APRT Study Group. Adenine phosphoribosyltransferase deficiency in children. *Pediatr Nephrol.* 2012 Apr;27(4):571-9. doi: 10.1007/s00467-011-2037-0. Epub 2012 Jan 3.
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