

DISEASE:
Adenine phosphoribosyltransferase deficiency

NAME:	Adenine phosphoribosyltransferase deficiency
DESCRIPTION:	A rare genetic nephropathy secondary to a disorder of purine metabolism characterized by the formation and hyperexcretion of 2,8-dihydroxyadenine (2,8-DHA) in urine, causing urolithiasis and crystalline nephropathy.
ORPHACODE:	976
SYNONYMS:	2,8-dihydroxyadenine urolithiasis APRT deficiency
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>APRT</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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