Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
2,8-dihydroxyadenine urolithiasis
Kamica 2,8 dihydroksyadeninowa
Niedobór APRT
Niedobór fosforybozylotransferazy adeninowej
APRT deficiency

Kod ORPHA
976

Kod OMIM
614723

Kod ICD10
E79.8

Kod ICD11
5C55.0Y

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*Źródło

orphanet