

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

*Źródło

orphanet