

Opis choroby *

Definicja

A rare purine metabolism disorder due to inherited deficiency of the xanthine dehydrogenase/oxidase enzyme and is characterized by very low (or undetectable) concentrations of uric acid in blood and urine and very high concentration of xanthine in urine, leading to urolithiasis.

Dane

Klasyfikacja

Choroba

Synonimy

Classic xanthinuria
Choroba kamieni ksantynowych
Kamica ksantynowa
Ksantynuria klasyczna
Xanthic urolithiasis
Xanthine stone disease

Kod ORPHA

3467

Kod OMIM

603592

Kod ICD10

E79.8

Kod ICD11

5C55.00

*Źródło

orphanet