Opis choroby *

Definicja

Type I xanthinuria, a type of classical xanthinuria (see this term), is a rare autosomal recessive disorder of purine metabolism (see this term) characterized by the isolated deficiency of xanthine dehydrogenase, causing hyperxanthinemia with low or absent uric acid and xanthinuria, leading to urolithiasis, hematuria, renal colic and urinary tract infections, while some patients are asymptomatic and others suffer from kidney failure. Less common manifestations include arthropathy, myopathy and duodenal ulcer.

Dane

Klasyfikacja
Podtyp etiologiczny

Synonimy
XDH deficiency
Niedobór dehydrogenazy ksantynowej
Niedobór oksydazy ksantynowej
Niedobór oksydoreduktazy ksantynowej
Niedobór XDH
Niedobór XO
Niedobór XOR
XO deficiency
XOR deficiency
Xanthine dehydrogenase deficiency
Xanthine oxidase deficiency
Xanthine oxidoreductase deficiency

Kod ORPHA
93601

Kod OMIM
278300

Kod ICD10
E79.8

Kod ICD11
5C55.00

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*Źródło

orphanet