Opis choroby *

Definicja

Type II xanthinuria, a type of classical xanthinuria (see this term), is a rare autosomal recessive disorder of purine metabolism (see this term) characterized by the deficiency of both xanthine dehydrogenase and aldehyde oxidase, leading to the formation of urinary xanthine urolithiasis and leading, in some patients, to kidney failure. Other less common manifestations include arthropathy, myopathy and duodenal ulcer, while some patients remain asymptomatic.

Dane

Klasyfikacja
Podtyp etiologiczny

Synonimy
XDH and AOX dual deficiency
Podwójny Niedobór dehydrogenazy ksantynowej i oksydazy aldehydowej ksantyny
Podwójny Niedobór XDH i AOX
Xanthine dehydrogenase and xanthine aldehyde oxidase dual deficiency

Kod ORPHA
93602

Kod OMIM
603592

Kod ICD10
E79.8

Kod ICD11
5C55.00

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*Źródło

orphanet