

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

*Źródło

orphanet