Xanthinuria type II

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Xanthinuria type II. ORPHA:93602

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.