Hereditary xanthinuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hereditary xanthinuria. ORPHA:3467

Hereditary xanthinuria is a 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.