Primary hyperoxaluria type 1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Primary hyperoxaluria type 1. ORPHA:93598

Primary hyperoxaluria type 1 (PH1) is a rare disorder of glyoxylate metabolism characterized by the accumulation of oxalate due to a deficiency of the peroxisomal hepatic enzyme L-alanine: glyoxylate aminotransferase (AGT). Clinical presentation is variable, ranging from occasional symptomatic nephrolithiasis to nephrocalcinosis and end-stage renal disease with systemic involvement.