DISEASE:
Primary hyperoxaluria type 1

NAME:	Primary hyperoxaluria type 1
DESCRIPTION:	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.
ORPHACODE:	93598
SYNONYMS:	Glycolic aciduria Peroxisomal alanine-glyoxylate aminotransferase deficiency
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	AGXT
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RELATED CONTENT

Related Genetic Tests

- Hyperoxaluria
- Tubulopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- alanine--glyoxylate aminotransferase

Related Gene Panels

- Tubulopathy/Nephrolithiasis (106 genes) - IPG