DISEASE:
Infantile hypophosphatasia

NAME:	Infantile hypophosphatasia
DESCRIPTION:	A rare, severe, genetic form of hypophosphatasia (HPP) characterized by infantile rickets without elevated serum alkaline phosphatase (ALP) activity and a wide range of clinical manifestations due to hypomineralization.
ORPHACODE:	247651
SYNONYMS:	Infantile Rathbun disease Infantile phosphoethanolaminuria
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ALPL
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Genetic disorders of Calcium and Phosphate metabolism (gene panel)
- Hypophosphatasia
- Tubulopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- alkaline phosphatase, biomineralization associated

Related Gene Panels

- Genetic disorders of Calcium and Phosphate metabolism (31 genes) - KUL
- Tubulopathy/Nephrolithiasis (106 genes) - IPG