DISEASE:
Childhood-onset hypophosphatasia

NAME:	Childhood-onset hypophosphatasia
DESCRIPTION:	A rare, moderate form of hypophosphatasia (HPP) characterized by onset after six months of age and widely variable clinical features from low bone mineral density for age, to unexplained fractures, skeletal deformities, and rickets with short stature and waddling gait.
ORPHACODE:	247667
SYNONYMS:	Childhood-onset Rathbun disease Childhood-onset phosphoethanolaminuria
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ALPL
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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