DISEASE:
Perinatal lethal hypophosphatasia

NAME:	Perinatal lethal hypophosphatasia
DESCRIPTION:	A rare, genetic form of hypophosphatasia (HPP) characterized by markedly impaired bone mineralization in utero due to reduced activity of serum alkaline phosphatase (ALP) and causing stillbirth or respiratory failure within days of birth.
ORPHACODE:	247623
SYNONYMS:	Perinatal lethal Rathbun disease Perinatal lethal 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

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