DISEASE:
Hereditary hypophosphatemic rickets with hypercalciuria

NAME:	Hereditary hypophosphatemic rickets with hypercalciuria
DESCRIPTION:	A rare hereditary disorder of renal phosphate wasting characterized by hypophosphatemia and hypercalciuria associated with rickets and/or osteomalacia. Other features include slow growth, short stature, skeletal deformities, muscle weakness and bone pain that are associated with normal or elevated plasma levels of calcitriol and hyperphosphaturia.
ORPHACODE:	157215
SYNONYMS:	HHRH
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SLC34A3 SLC34A1
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RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

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

Related Analytes

- solute carrier family 34 member 1
- solute carrier family 34 member 3

Related Gene Panels

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