DISEASE:
Dominant hypophosphatemia with nephrolithiasis or osteoporosis

NAME:	Dominant hypophosphatemia with nephrolithiasis or osteoporosis
DESCRIPTION:	A rare, genetic renal tubular disease characterized by phosphate loss in the proximal tubule, leading to hypercalciuria and recurrent urolithiasis and/or osteoporosis.
ORPHACODE:	244305
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	SLC34A1 NHERF1
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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

- NHERF family PDZ scaffold protein 1
- solute carrier family 34 member 1

Related Gene Panels

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