DISEASE:
Autosomal recessive hypophosphatemic rickets

NAME:	Autosomal recessive hypophosphatemic rickets
DESCRIPTION:	A rare, autosomal recessive renal phosphate-wasting disorder characterized by childhood-onset hypophosphatemia that clinically manifests with rickets and/or osteomalacia, slow growth/short stature, bone pain and skeletal deformities. Additional findings may include fatigue, muscle weakness and repeated bone fractures.
ORPHACODE:	289176
SYNONYMS:	ARHR
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	DMP1 ENPP1
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)
- Tubulopathy (gene panel)

Related Laboratories

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

Related Analytes

- dentin matrix acidic phosphoprotein 1
- ectonucleotide pyrophosphatase/phosphodiesterase 1

Related Gene Panels

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

Source URL: http://gentest.healthdata.be/disease/1988