DISEASE:
Primary Fanconi renotubular syndrome

NAME:	Primary Fanconi renotubular syndrome
DESCRIPTION:	A rare generalized, genetic disorder of proximal tubular transport characterized by excessive urine output with loss of low molecular weight solutes (amino acids, glucose, low-molecular weight proteins, organic acids, carnitine, calcium, phosphate, potassium, bicarbonate) and water, and which can be life threatening.
ORPHACODE:	3337
SYNONYMS:	DeToni-Debré-Fanconi syndrome Primary Fanconi renal syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM
ANALYTE(S):	EHHADH GATM SLC34A1 NDUFAF6
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RELATED CONTENT

Related Genetic Tests

- Hepatorenal disorders (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)
- Tubulopathy (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- enoyl-CoA hydratase and 3-hydroxyacyl CoA dehydrogenase
- glycine amidinotransferase
- NADH:ubiquinone oxidoreductase complex assembly factor 6
- solute carrier family 34 member 1

Related Gene Panels

- Tubulopathy/Nephrolithiasis (106 genes) - IPG