DISEASE:
Autosomal recessive infantile hypercalcemia

NAME:	Autosomal recessive infantile hypercalcemia
DESCRIPTION:	A rare, genetic, phosphocalcic metabolism disorder characterized by early-onset hypercalcemia, hypophosphatemia, hypercalciuria, decreased intact parathyroid hormone serum levels and medullary nephrocalcinosis, typically manifesting with failure to thrive, hypotonia, vomiting, constipation and/or polyuria.
ORPHACODE:	300547
SYNONYMS:	Familial infantile hypercalcemia with suppressed intact parathyroid hormone
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	CYP24A1 SLC34A1
CREATED:	13 May 2019 - 01:02
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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

- cytochrome P450 family 24 subfamily A member 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