DISEASE:
Familial hypercholanemia

NAME:	Familial hypercholanemia
DESCRIPTION:	Familial hypercholanemia is a very rare genetic disorder characterized clinically by elevated serum bile acid concentrations, itching, and fat malabsorption reported in patients of Old Order Amish descent.
ORPHACODE:	238475
SYNONYMS:	Hereditary hypercholanemia
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10
ANALYTE(S):	EPHX1 TJP2 BAAT SLC10A1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1608

RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Familial hypercholanemia (3 genes)

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- bile acid-CoA:amino acid N-acyltransferase
- epoxide hydrolase 1
- solute carrier family 10 member 1
- tight junction protein 2

Related Gene Panels

- Cholestasis (40 genes) - UCL
- Familial hypercholanemia (3 genes) - UCL