DISEASE:
Sitosterolemia

NAME:	Sitosterolemia
DESCRIPTION:	Sitosterolemia is a rare autosomal recessive sterol storage disease characterized by the accumulation of phytosterols in the blood and tissues. Clinical manifestations include xanthomas, arthralgia and premature atherosclerosis. Hematological manifestations include hemolytic anemia with stomatocytosis and macrothrombocytopenia. The disease is caused by homozygous or compound heterozygous mutations in ABCG5 (2p21) and ABCG8 (2p21) genes.
ORPHACODE:	2882
SYNONYMS:	Phytosterolemia
XREF(S):	Orphanet MeSH OMIM OMIM MedDRA ICD-10
ANALYTE(S):	ABCG5 ABCG8
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RELATED CONTENT

Related Genetic Tests

- Dyslipidemia (gene panel)
- Hypercholesterolemia, Familial (9 genes)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily G member 5
- ATP binding cassette subfamily G member 8

Related Gene Panels

- Dyslipidemia (13 genes) - ULB
- Familial Hypercholesterolemia panel (9 genes) - ULG
- Trombosis - Hemostasis (107 genes) - KUL