DISEASE:
Homozygous familial hypercholesterolemia

NAME:	Homozygous familial hypercholesterolemia
DESCRIPTION:	A rare disorder of lipid metabolism characterized by severely elevated plasma total cholesterol, low-density lipoprotein (LDL) cholesterol levels, and subsequent premature formation of atherosclerotic plaques in the coronary arteries, proximal aorta, and other arteries, significantly increasing the risk of premature cardiovascular disease and death. Xanthomas of the skin and in tendons are also a hallmark of the disease. Lethality is high due to early complications, in particular myocardial infarction and aortic valvular disease.
ORPHACODE:	391665
SYNONYMS:	HoFH
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10 OMIM

ANALYTE(S):	ABCG5 ABCG8 APOB LDLRAP1 PCSK9 LDLR
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RELATED CONTENT

Related Genetic Tests

- Dyslipidemia (gene panel)
- Hypercholesterolemia (9 genes)
- Hypercholesterolemia, Familial (4 genes)
- Hypercholesterolemia, Familial (9 genes)
- Hypercholesterolemia, Familial (Gene Panel)
- Hypercholesterolemia, Familial (gene panel)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily G member 5
- ATP binding cassette subfamily G member 8
- apolipoprotein B
- low density lipoprotein receptor

- low density lipoprotein receptor adaptor protein 1
- proprotein convertase subtilisin/kexin type 9

Related Gene Panels

- Dyslipidemia (13 genes) - ULB
- Familial Hypercholesterolemia (9 genes) - IPG
- Familial Hypercholesterolemia panel (8 genes) - UZA
- Familial Hypercholesterolemia panel (9 genes) - ULG
- Hypercholesterolemia (4 genes) - UZA
- Hypercholesterolemia (9 genes) - UCL
- Hypercholesterolemia, Familial (4 genes) - KUL
- Trombosis - Hemostasis (107 genes) - KUL
