DISEASE:
Cholesteryl ester storage disease

NAME:	Cholesteryl ester storage disease
DESCRIPTION:	A form of lysosomal acid lipase deficiency characterized by progressive cholesterol esters and triglyceride accumulation in tissues and organs typically presenting with hepatosplenomegaly, liver dysfunction and/or dyslipidemia.
ORPHACODE:	75234
SYNONYMS:	Cholesterol ester storage disease
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	LIPA
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RELATED CONTENT

Related Genetic Tests

- Dyslipidemia (gene panel)
- Hypercholesterolemia, Familial (9 genes)
- Hypercholesterolemia, Familial (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- lipase A, lysosomal acid type

Related Gene Panels

- Dyslipidemia (13 genes) - ULB
- Familial Hypercholesterolemia panel (8 genes) - UZA
- Familial Hypercholesterolemia panel (9 genes) - ULG

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