Belgian Genetic Tests database

DISEASE:
GRACILE syndrome

NAME:	GRACILE syndrome
DESCRIPTION:	An inherited lethal mitochondrial disorder characterized by fetal growth restriction (GR), aminoaciduria (A), cholestasis (C), iron overload (I), lactacidosis (L), and early death (E).
ORPHACODE:	53693
SYNONYMS:	Fellman disease Growth restriction-aminoaciduria-cholestasis-iron overload-lactic acidosis-early death syndrome
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	BCS1L
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Source URL: http://gentest.healthdata.be/disease/3006

RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Hepatorenal disorders (gene panel)
- Mitochondrial complex III deficiency / Bjornstad syndrome / Gracile syndrome
- Mitochondrial disorders (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone

Related Gene Panels

- Cholestasis (40 genes) - UCL
- mitochondrial disease, nuclear based (343 genes) - VUB