DISEASE:
Congenital bile acid synthesis defect type 2

NAME:	Congenital bile acid synthesis defect type 2
DESCRIPTION:	Congenital bile acid synthesis defect type 2 (BAS defect type 2) is an anomaly of bile acid synthesis (see this term) characterized by severe and rapidly progressive cholestatic liver disease, and malabsorption of fat and fat-soluble vitamins.
ORPHACODE:	79303
SYNONYMS:	BASD2 Cholestasis with delta(4)-3-oxosteroid 5-beta-reductase deficiency
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	AKR1D1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Bile Acid Synthesis Congenital Defect (gene panel)
- Cholestasis (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- aldo-keto reductase family 1 member D1

Related Gene Panels

- Cholestasis (40 genes) - UCL