DISEASE:
Congenital bile acid synthesis defect type 3

NAME:	Congenital bile acid synthesis defect type 3
DESCRIPTION:	Congenital bile acid synthesis defect type 3 (BAS defect type 3) is a severe anomaly of bile acid synthesis (see this term) characterized by severe neonatal cholestatic liver disease.
ORPHACODE:	79302
SYNONYMS:	BASD3 Oxysterol 7-alpha-hydroxylase deficiency
XREF(S):	Orphanet ICD-10 MeSH OMIM
ANALYTE(S):	CYP7B1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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