DISEASE:
Congenital bile acid synthesis defect type 4

NAME:	Congenital bile acid synthesis defect type 4
DESCRIPTION:	Congenital bile acid synthesis defect type 4 (BAS defect type 4) is an anomaly of bile acid synthesis (see this term) characterized by mild cholestatic liver disease, fat malabsorption and/or neurological disease.
ORPHACODE:	79095
SYNONYMS:	2-methylacyl-CoA racemase deficiency AMACR deficiency Alpha-methyl-acyl-CoA racemase deficiency BASD4 Liver disease-retinitis pigmentosa-polyneuropathy-epilepsy syndrome
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	AMACR
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