DISEASE:
Abetalipoproteinemia

NAME:	Abetalipoproteinemia
DESCRIPTION:	A severe, familial hypobetalipoproteinemia characterized by permanently low levels (below the 5th percentile) of apolipoprotein B and LDL cholesterol, and by growth delay, malabsorption, hepatomegaly, and neurological and neuromuscular manifestations.
ORPHACODE:	14
SYNONYMS:	Bassen-Kornzweig disease Homozygous familial hypobetalipoproteinemia
XREF(S):	Orphanet MeSH OMIM ICD-10 OMIM OMIM
ANALYTE(S):	MTTP
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