DISEASE:
Holocarboxylase synthetase deficiency

NAME:	Holocarboxylase synthetase deficiency
DESCRIPTION:	A rare, early-onset and life-threatening, multiple carboxylase deficiency that when left untreated, is characterized by vomiting, tachypnea, irritability, lethargy, exfoliative dermatitis, and seizures that can worsen to coma and death.
ORPHACODE:	79242
SYNONYMS:	Early-onset multiple carboxylase deficiency Neonatal multiple carboxylase deficiency
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	HLCS
CREATED:	13 May 2019 - 01:02
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