

DISEASE:
3-methylcrotonyl-CoA carboxylase deficiency

NAME:	3-methylcrotonyl-CoA carboxylase deficiency
DESCRIPTION:	A rare inherited disorder of leucine metabolism characterized by a highly variable clinical picture ranging from metabolic crisis in infancy to asymptomatic adults.
ORPHACODE:	6
SYNONYMS:	3-methylcrotonylglycinuria MCC deficiency MCCD
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	MCCC1 MCCC2
CREATED:	13 May 2019 - 01:02
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Source URL: <http://gentest.healthdata.be/disease/1310>

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