DISEASE:
Vitamin B12-unresponsive methylmalonic acidemia type mut-

NAME:	Vitamin B12-unresponsive methylmalonic acidemia type mut-
DESCRIPTION:	Vitamin B12-unresponsive methylmalonic acidemia type mut- is an inborn error of metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12.
ORPHACODE:	79312
SYNONYMS:	Partial deficiency of methylmalonyl-CoA mutase Vitamin B12-unresponsive methylmalonic aciduria type mut-
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MMUT
CREATED:	13 May 2019 - 01:02
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