DISEASE:
Vitamin B12-unresponsive methylmalonic acidemia type mut0

NAME:	Vitamin B12-unresponsive methylmalonic acidemia type mut0
DESCRIPTION:	Vitamin B12-unresponsive methylmalonic acidemia type mut0 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:	289916
SYNONYMS:	Complete deficiency of methylmalonyl-CoA mutase Vitamin B12-unresponsive methylmalonic aciduria type mut0
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MMUT
CREATED:	13 May 2019 - 01:02
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