

DISEASE:**Methylmalonic aciduria due to transcobalamin receptor defect**

NAME:	Methylmalonic aciduria due to transcobalamin receptor defect
DESCRIPTION:	Methylmalonic aciduria due to transcobalamin receptor defect is a rare metabolite absorption and transport disorder characterized by a moderate increase of methylmalonic acid (MMA) in the blood and urine due to decreased cellular uptake of cobalamin resulting from decreased transcobalamin receptor function. Patients are usually asymptomatic however, screening reveals increased C3-acylcarnitine and MMA in plasma. Serum homocysteine levels may vary from normal to moderately elevated and retinal vascular occlusive disease, resulting in severe visual loss, has been reported.
ORPHACODE:	280183
SYNONYMS:	Methylmalonic acidemia, TCb1R type Methylmalonic acidemia, TCb1R type
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CD320
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