

Opis choroby *

Definicja

Methylmalonic acidemia is an inborn error of vitamin B12 metabolism characterized by gastrointestinal and neurometabolic manifestations resulting from decreased function of the mitochondrial enzyme methylmalonyl-CoA mutase.

Dane

Klasyfikacja	Synonimy
Grupa fenomenów	Methylmalonic aciduria without homocystinuria Acyduria metylomalonowa bez homocystynurii

Kod ORPHA	Kod OMIM	Kod ICD10
293355	-	E71.1

Kod ICD11
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*Źródło

orphanet