Methylmalonic acidemia without homocystinuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Methylmalonic acidemia without homocystinuria. ORPHA:293355

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.