Methylmalonic acidemia with homocystinuria, type cblC

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Methylmalonic acidemia with homocystinuria, type cblC. ORPHA:79282

cblC type methylmalonic acidemia with homocystinuria is a form of methylmalonic acidemia with homocystinuria (see this term), an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures.