Methylmalonic acidemia with homocystinuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Methylmalonic acidemia with homocystinuria. ORPHA:26

Methylmalonic acidemia with homocystinuria is an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures. There are four complementation classes of cobalamin defects (cblC, cblD, cblF and cblJ) that are responsible for methylmalonic acidemia - homocystinuria (methylmalonic acidemia - homocystinuria cblC, cblD cblF and cblJ; see these terms).