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Vitamin B12-unresponsive methylmalonic acidemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Vitamin B12-unresponsive methylmalonic acidemia. ORPHA:27

Vitamin B12-unresponsive methylmalonic acidemia is an inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic crises or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12. There are two types of vitamin B12-unresponsive methylmalonic acidemia: mut0 and mut- (see these terms).