3-hydroxy-3-methylglutaric aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3-hydroxy-3-methylglutaric aciduria. ORPHA:20

3-hydroxy-3-methylglutaric aciduria (3HMG) is an organic aciduria, due to deficiency of 3-hydroxy-3-methylglutaryl-CoA-lyase (a key enzyme in ketogenesis and leucine metabolism) usually presenting in infancy with episodes of metabolic decompensation triggered by periods of fasting or infections, which when left untreated are life-threatening and may lead to neurological sequelae.