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Intermittent maple syrup urine disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Intermittent maple syrup urine disease. ORPHA:268173

Intermittent maple syrup urine disease (intermittent MSUD) is a mild form of MSUD (see this term) where patients (when well) are asymptomatic with normal levels of branched-chain amino acids (BCAAs) but with catabolic stress are at risk of acute decompensation with ketoacidosis, which can lead to cerebral edema and coma if untreated.