Maple syrup urine disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Maple syrup urine disease. ORPHA:511

Maple syrup urine disease (MSUD) is a rare inherited disorder of branched-chain amino acid metabolism classically characterized by poor feeding, lethargy, vomiting and a maple syrup odor in the cerumen (and later in urine) noted soon after birth, followed by progressive encephalopathy and central respiratory failure if untreated. The four overlapping phenotypic subtypes are: classic, intermediate, intermittent and thiamine-responsive MSUD (see these terms).