Open Peer Review on Qeios

Classic maple syrup urine disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Classic</u> <u>maple syrup urine disease</u>. ORPHA:268145

Classic maple syrup urine disease (classic MSUD) is the most severe and probably common form of MSUD (see this term) characterized by a maple syrup odor in the cerumen at birth, poor feeding, lethargy and focal dystonia, followed by progressive encephalopathy and central respiratory failure if untreated.