Isovaleric acidemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Isovaleric acidemia. ORPHA:33

Isovaleric acidemia (IVA) is an autosomal recessively inherited organic aciduria characterized by a deficiency in isovaleryl-CoA dehydrogenase, that has wide clinical variability and that can present in infancy with acute manifestations of vomiting, failure to thrive, seizures, lethargy, a characteristic ''sweaty feet'' odor, acute pancreatitis and mild to severe developmental delay or in childhood with metabolic acidosis (brought on by prolonged fasting, an increased intake of protein-rich food or infections) and that can be fatal if not treated immediately. Chronic intermittent presentations and asymptomatic patients have also been reported.