Isobutyryl-CoA dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Isobutyryl-CoA dehydrogenase deficiency. ORPHA:79159

Isobutyryl-CoA dehydrogenase deficiency is an inborn error of valine metabolism. The prevalence is unknown. Only one symptomatic patient (with anaemia, failure to thrive, dilated cardiomyopathy and plasma carnitine deficiency) has been described so far, but several series of patients have been identified through newborn screening programs relying on detection of increased C(4)-carnitine levels by tandem mass spectrometry. The disorder is caused by mutations in the ACAD8 gene (11q25).