3-hydroxyisobutyric aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3-hydroxyisobutyric aciduria. ORPHA:939

3 hydroxyisobutyric aciduria is characterised by ketoacidotic episodes, cerebral anomalies and facial dysmorphism. It is an organic aciduria that involves valine metabolism. Thirteen cases have been described in the literature so far. Transmission is thought to be autosomal recessive.