Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency. ORPHA:88639

Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency is characterised by delayed motor development, hypotonia and progressive neurodegeneration. To date, it has been described in four boys. The syndrome is caused by mutations affecting the two alleles of the HIBCH gene, encoding 3-hydroxyisobutyryl-CoA hydrolase. The mode of transmission has not yet been established.