Hydroxykynureninuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hydroxykynureninuria. ORPHA:79155

Encephalopathy due to hydroxykynureninuria is characterised by psychomotor retardation and nonprogressive encephalopathy associated with urinary excretion of large amounts of kynurenine, 3-hydroxykynurenine, and xanthurenic acid. It has been described in less than 30 patients. Other manifestations may include muscular hypertonia, headaches and stereotyped gestures. This disorder is transmitted as an autosomal recessive trait. It is caused by a defect in kynureninase, an enzyme of the tryptophane catabolic pathway.