Citrullinemia type I

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Citrullinemia type I. ORPHA:247525

Citrullinemia type I is a rare autosomal recessive urea cycle defect characterized biologically by hyperammonemia and clinically by progressive lethargy, poor feeding and vomiting in the neonatal form (Acute neonatal citrullinemia type I, see this term) and by variable hyperammonemia in the later-onset form (Adult-onset citrullinemia type I, see this term).