Citrin deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Citrin deficiency. ORPHA:247582

Citrin deficiency is a rare autosomal recessive urea cycle defect characterized clinically by recurring episodes of hyperammonemia and associated neuropsychiatric symptoms in the adult-onset form (citrullinemia type II, see this term), and by transient cholestasis and variable hepatic dysfunction in the neonatal form (neonatal intrahepatic cholestasis due to citrin deficiency, see this term).