Carbamoyl-phosphate synthetase 1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Carbamoyl-phosphate synthetase 1 deficiency. ORPHA:147

Carbamoyl-phosphate synthetase 1 deficiency (CPS1D) is a rare and severe disorder of urea cycle metabolism most commonly characterized by either a neonatal-onset of severe hyperammonemia that occurs few days after birth and manifests with lethargy, vomiting, hypothermia, seizures, coma and death or a presentation outside the newborn period at any age with (sometimes) milder symptoms of hyperammonemia.