

Opis choroby *

Definicja

A rare, severe disorder of urea cycle metabolism typically 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.

Dane

Klasyfikacja	Synonimy
Choroba	CPS1 deficiency
	CPS1D
	Niedobór CPS1
	Niedobór syntetazy karbamoilofosforanowej
	Niedobór syntetazy karbamoilofosforanowej 1
	CPS1D
	Carbamoyl-phosphate synthetase I deficiency
	Carbamoyl-phosphate synthetase deficiency

Kod ORPHA
147

Kod OMIM
237300

Kod ICD10
E72.2

Kod ICD11
5C50.A1

*Źródło

orphanet