

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

Choroba

Synonimy

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