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

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*Źródło

orphanet