Opis choroby *

Definicja

A rare, genetic disorder of urea cycle metabolism typically characterized by either a severe, neonatal-onset form that manifests with hyperammonemia accompanied with vomiting, hypothermia, lethargy and poor feeding in the first few days of life, or late-onset forms that manifest with stress- or infection-induced episodic hyperammonemia or, in some, behavioral abnormalities and/or learning disabilities, or chronic liver disease. Patients often manifest liver dysfunction.

Dane

Klasyfikacja
Choroba

Synonimy
ASA deficiency
Niedobór argininosukcynazy
Niedobór ASA
Niedobór ASL
Niedobór liazy argininobursztynianowej
Niedobór liazy kwasu argininobursztynowego
ASL deficiency
Argininosuccinase deficiency
Argininosuccinatelyase deficiency
Argininosuccinic acid lyase deficiency

Kod ORPHA
23

Kod OMIM
207900

Kod ICD10
E72.2

Kod ICD11
5C50.A0

---

*Źródło

orphanet