## Opis choroby *

Definicja

Citrullinemia type I is a rare autosomal recessive urea cycle defect characterized biologically by hyperammonemia and clinically by progressive lethargy, poor feeding and vomiting in the neonatal form (Acute neonatal citrullinemia type I, see this term) and by variable hyperammonemia in the later-onset form (Adult-onset citrullinemia type I, see this term).

Dane

**Klasyfikacja**
Choroba

Synonimy
ASS deficiency
Citrulinemia klasyczna
Citrulinemia typu 1
CTLN1
Niedobór ASS
Niedobór syntazy argininobursztynianu
Niedobór syntazy kwasu argininobursztynowego
Niedobór syntetazy argininobursztynianu
Niedobór syntetazy kwasu argininobursztynowego
Argininosuccinate synthase deficiency
Argininosuccinate synthetase deficiency
Argininosuccinic acid synthase deficiency
Argininosuccinic acid synthetase deficiency
CTLN1
Citrullinemia type 1
Classic citrullinemia

**Kod ORPHA**
247525

**Kod OMIM**
215700

**Kod ICD10**
E72.2

**Kod ICD11**
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*Źródło

orphanet