## Opis choroby *

Definicja

A rare, genetic disorder of urea cycle metabolism and ammonia detoxification characterized by either a severe, neonatal-onset disease found mainly in males, or later-onset (partial) forms of the disease. Both present with episodes of hyperammonemia that can be fatal and which can lead to neurological sequelae.

Dane

**Klasyfikacja**
Choroba

Synonimy
OCT deficiency
Niedobór karbamylotransferazy ornityny
Niedobór OCT
Niedobór OTC
OTC deficiency
Ornithine carbamoyltransferase deficiency

**Kod ORPHA**
664

**Kod OMIM**
311250

**Kod ICD10**
E72.4

**Kod ICD11**
5C50.AY

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*Źródło

orphanet