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