

Opis choroby *

Definicja

A severe subtype of citrin deficiency characterized clinically by adult onset (20 and 50 years of age), recurrent episodes of hyperammonemia and associated neuropsychiatric symptoms such as nocturnal delirium, confusion, restlessness, disorientation, drowsiness, memory loss, abnormal behavior (aggression, irritability, and hyperactivity), seizures, and coma.

Dane

Klasyfikacja

Choroba

Synonimy

Adult-onset citrin deficiency
Citrulinemia typu 2
Citrulinemia typu 2 o początku w wieku dorosłym
Citrulinemia typu II o początku w wieku dorosłym
CTLN2
Niedobór cytrynianu o początku w wieku dorosłym
Adult-onset citrullinemia type 2
Adult-onset citrullinemia type II
CTLN2
Citrullinemia type 2

Kod ORPHA

247585

Kod OMIM

603471

Kod ICD10

E72.2

Kod ICD11

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*Źródło

orphanet