

## 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