

## Opis choroby \*

### Definicja

A severe form of citrullinemia type 1 characterized biologically by hyperammonemia and clinically by progressive lethargy, poor feeding and vomiting, seizures and possible loss of consciousness, within one to a few days of birth, with variable signs of increased intracranial pressure. The condition can lead to significant neurologic deficits.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Acute neonatal citrullinemia type 1  
Citrulinemia klasyczna typu 1  
Citrulinemia klasyczna typu I  
Ostra citrulinemia noworodkowa typu 1  
Ostra noworodkowa cytrulinemia typu I  
Early-onset citrullinemia type 1  
Early-onset citrullinemia type I

#### Kod ORPHA

247546

#### Kod OMIM

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#### Kod ICD10

E72.2

#### Kod ICD11

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#### \*Źródło

orphanet