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