Opis choroby *

Definicja

Neonatal glycine encephalopathy is a frequent, usually severe form of glycine encephalopathy (GE; see this term) characterized by coma, apnea, hypotonia, seizure and myoclonic jerks in the neonatal period, and subsequent developmental delay.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Classic glycine encephalopathy
Klasyczna encefalopatia glicynowa
Nieketonowa hiperglicynemia noworodków
NKH noworodków
Neonatal NKH
Neonatal non-ketotic hyperglycinemia

Kod ORPHA
289857

Kod OMIM
605899

Kod ICD10
E72.5

Kod ICD11
5C50.70

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*Źródło

orphanet