

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

*Źródło

orphanet