

Opis choroby *

Definicja

A very rare neonatal epileptic encephalopathy disorder characterized clinically by onset of severe seizures within hours of birth that are not responsive to anticonvulsants, but are responsive to treatment with pyridoxal phosphate.

Dane

Klasyfikacja

Choroba

Synonimy

PNPO deficiency

Napady zależne od fosforanu pirydoksalu

Niedobór 5'-oksydazy pirydoksaminy

Niedobór oksydazy 5'-fosforanu pirydoksyaminy

Niedobór PNPO

Noworodkowa encefalopatia epileptyczna

związana z PNPO

PNPO-related neonatal epileptic encephalopathy

Pyridoxal phosphate-dependent seizures

Pyridoxamine 5'-oxidase deficiency

Pyridoxamine 5'-phosphate oxidase deficiency

Kod ORPHA

79096

Kod OMIM

610090

Kod ICD10

G40.8

Kod ICD11

8A61.00

*Źródło

orphanet