

## 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 pirydoksaminy

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

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#### \*Źródło

orphanet