

Opis choroby *

Definicja

A rare neurometabolic disease characterized by recurrent intractable seizures in the prenatal, neonatal and postnatal period that are resistant to anti-epileptic drugs (AEDs) but that are responsive to pharmacological dosages of pyridoxine (vitamin B6).

Dane

Klasyfikacja

Choroba

Synonimy

Antiquitin deficiency

Napady padaczkowe zależne od witaminy B6

Niedobór antykwityny

Vitamin B6-dependent seizures

Kod ORPHA

3006

Kod OMIM

266100

Kod ICD10

G40.8

Kod ICD11

8A61.0Y

*Źródło

orphanet