

Opis choroby *

Definicja

Benign familial neonatal-infantile seizures (BFNIS) is a benign familial epilepsy syndrome with an intermediate phenotype between benign familial neonatal seizures (BFNS) and benign familial infantile seizures (BFIS; see these terms). So far, this syndrome has been described in multiple members of 10 families. Age of onset in these BFNIS families varied from 2 days to 6 months, with spontaneous resolution in most cases before the age of 12 months. Like BFNS and BFIS, seizures in BFNIS generally occur in clusters over one or a few days with posterior focal seizure onset. BFNIS is caused by mutations in the *SCN2A* gene (2q24.3), encoding the voltage-gated sodium channel alpha-subunit Na(V)1.2. Transmission is autosomal dominant.

Dane

Klasyfikacja

Choroba

Synonimy

BFNIS

BFNIS

Łagodna padaczka noworodkowo-dziecięca

Benign neonatal-infantile epilepsy

Kod ORPHA

140927

Kod OMIM

607745

Kod ICD10

G40.4

Kod ICD11

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[*Źródło](#)

orphanet