Benign familial neonatal-infantile seizures

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Benign familial neonatal-infantile seizures. ORPHA:140927

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.