DISEASE:
Benign familial neonatal-infantile seizures

NAME:	Benign familial neonatal-infantile seizures
DESCRIPTION:	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.
ORPHACODE:	140927
SYNONYMS:	BFNIS Benign neonatal-infantile epilepsy
XREF(S):	Orphanet OMIM MedDRA ICD-10
ANALYTE(S):	SCN2A KCNQ2

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/739

RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- potassium voltage-gated channel subfamily Q member 2
- sodium voltage-gated channel alpha subunit 2

Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA
