DISEASE:
Non-specific early-onset epileptic encephalopathy

NAME:	Non-specific early-onset epileptic encephalopathy
DESCRIPTION:	A rare infantile epilepsy syndrome characterized by early onset of seizures of variable type and severity, potentially associated with a spectrum of clinical signs and symptoms including delay or lack of psychomotor development, intellectual disability, poor or absent speech development, behavioral abnormalities, hypotonia, movement disorders, spasticity, microcephaly, and dysmorphic facial features, among others. Brain imaging findings are also variable and may include cerebral atrophy or white matter abnormalities.
ORPHACODE:	442835
SYNONYMS:	Non-specific EOEE Undetermined EOEE Undetermined early-onset epileptic encephalopathy

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	ACTL6B GABRG2 GABRA2 FZR1 GABRA5 PACS2 YWHAG UBA5 GRIN2D DALRD3 TRAK1 SLC1A2 AP3B2 ATP6V1A SCN3A GABRB2 CLTC NUS1 CYFIP2 FBXO28 NTRK2 SLC38A3 FGF12 SZT2 FGF13 CELF2 DHDDS PPP3CA SYNJ1 SYNGAP1 WWOX AARS1 SCN8A NECAP1 HCN1 SLC13A5 KCNB1 DNM1 KCNA2

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

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- Epilepsy (gene panel)
- Epilepsy, seizures (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- alanyl-tRNA synthetase 1
- actin like 6B
- adaptor related protein complex 3 subunit beta 2
- ARV1 homolog, fatty acid homeostasis modulator
- ATPase Na+/K+ transporting subunit alpha 2
- ATPase Na+/K+ transporting subunit alpha 3
- ATPase H+ transporting V1 subunit A
- calcium voltage-gated channel subunit alpha1 A
- calcium voltage-gated channel subunit alpha1 B
- calcium voltage-gated channel auxiliary subunit alpha2delta 1
- cyclin dependent kinase 19

- CUGBP Elav-like family member 2
- clathrin heavy chain
- connector enhancer of kinase suppressor of Ras 2
- cytoplasmic FMR1 interacting protein 2
- DALR anticodon binding domain containing 3
- dehydrodolichyl diphosphate synthase subunit
- dynamin 1
- eukaryotic translation elongation factor 1 alpha 2
- F-box protein 28
- fibroblast growth factor 12
- fibroblast growth factor 13
- fizzy and cell division cycle 20 related 1
- gamma-aminobutyric acid type B receptor subunit 2
- gamma-aminobutyric acid type A receptor subunit alpha2
- gamma-aminobutyric acid type A receptor subunit alpha5
- gamma-aminobutyric acid type A receptor subunit beta2
- gamma-aminobutyric acid type A receptor subunit gamma2
- glutamate ionotropic receptor NMDA type subunit 2D
- hyperpolarization activated cyclic nucleotide gated potassium channel 1
- potassium voltage-gated channel subfamily A member 2
- potassium voltage-gated channel subfamily B member 1
- NECAP endocytosis associated 1
- neurotrophic receptor tyrosine kinase 2
- NUS1 dehydrodolichyl diphosphate synthase subunit
- phosphofurin acidic cluster sorting protein 2
- prolyl-tRNA synthetase 2, mitochondrial
- protein phosphatase 3 catalytic subunit alpha
- sodium voltage-gated channel alpha subunit 3
- sodium voltage-gated channel alpha subunit 8
- solute carrier family 13 member 5
- solute carrier family 1 member 2
- solute carrier family 38 member 3
- synaptic Ras GTPase activating protein 1

- synaptojanin 1
- SZT2 subunit of KICSTOR complex
- trafficking kinesin protein 1
- ubiquitin like modifier activating enzyme 5
- WW domain containing oxidoreductase
- tyrosine 3-monooxygenase/tryptophan 5-monooxygenase activation protein gamma

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Epilepsy, seizures (196 genes) - IPG
- Rare epilepsy with developmental delay (> 240 genes) - UZA
