DISEASE:
Early infantile epileptic encephalopathy

NAME:	Early infantile epileptic encephalopathy
DESCRIPTION:	A severe form of age-related epileptic encephalopathies characterized by the onset of tonic spasms within the first 3 months of life that can be generalized or lateralized, independent of the sleep cycle, and that can occur hundreds of times per day, leading to psychomotor impairment and death.
ORPHACODE:	1934
SYNONYMS:	EIEE Early infantile epileptic encephalopathy with suppression-bursts Ohtahara syndrome

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

ANALYTE(S):	DMXL2 GRIN1 SLC32A1 NEUROD2 GRM7 SCN2A CDKL5 ARX CASK PNKP GNAO1 PIGQ SIK1 SCN1B SLC25A22 KCNA1 TRIM8 PIGP
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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- aristaless related homeobox
- calcium/calmodulin dependent serine protein kinase
- cyclin dependent kinase like 5
- Dmx like 2
- G protein subunit alpha o1
- glutamate ionotropic receptor NMDA type subunit 1
- glutamate metabotropic receptor 7
- potassium voltage-gated channel subfamily A member 1
- neuronal differentiation 2
- phosphatidylinositol glycan anchor biosynthesis class P
- phosphatidylinositol glycan anchor biosynthesis class Q
- polynucleotide kinase 3'-phosphatase
- sodium voltage-gated channel beta subunit 1
- sodium voltage-gated channel alpha subunit 2
- salt inducible kinase 1

- solute carrier family 25 member 22
- solute carrier family 32 member 1
- tripartite motif containing 8

Related Gene Panels

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