DISEASE:
Hyperekplexia-epilepsy syndrome

NAME:	Hyperekplexia-epilepsy syndrome
DESCRIPTION:	A rare, X-linked, syndromic intellectual disability disease characterized by neonatal hypertonia which evolves to hypotonia and an exaggerated startle response (to sudden visual, auditory or tactile stimuli), followed by the development of early-onset, frequently refractory, tonic or myoclonic seizures. Progressive epileptic encephalopathy, intellectual disability, and psychomotor development arrest, with subsequent decline, may be additionally associated.
ORPHACODE:	163985
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ARHGEF9
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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Epilepsy gene panel
- Hyperekplexia (gene panel-6 genes)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- Cdc42 guanine nucleotide exchange factor 9

Related Gene Panels

- Epilepsy gene panel - VUB
- Hyperekplexia (6 genes) - IPG
- Rare epilepsy with developmental delay (> 240 genes) - UZA

Source URL: http://gentest.healthdata.be/disease/95