DISEASE:
Hereditary hyperekplexia

NAME:	Hereditary hyperekplexia
DESCRIPTION:	Hereditary hyperekplexia is a hereditary neurological disorder characterized by excessive startle responses.
ORPHACODE:	3197
SYNONYMS:	Congenital stiff man syndrome Familial startle disease Hereditary hyperexplexia Kok disease Stiff baby syndrome
XREF(S):	Orphanet OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	ATAD1 GLRA1 GLRB GPHN SLC6A5
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Hyperekplexia (3 genes)
- Hyperekplexia (gene panel-6 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- ATPase family AAA domain containing 1
- glycine receptor alpha 1
- glycine receptor beta
- gephyrin
- solute carrier family 6 member 5

Related Gene Panels

- Hyperekplexia (3 genes) - ULG
- Hyperekplexia (6 genes) - IPG