Hyperekplexia

Precision Panel

Overview

Hyperekplexia, also known as stiff baby syndrome or startle disease, is a rare hereditary neurological disease associated to a variety of gene mutations that affect the glycine receptor. This disorder is characterized by a triad of generalized stiffness while awake, nocturnal myoclonus and exaggerated startle reflex. These features often appear at birth alongside episodes of hypertonia or tonic spasms upon awakening. When severe, it may interfere with breathing causing irreversible brain damage. Hyperekplexia is usually misdiagnosed as a form epilepsy. It is inherited mostly as an autosomal dominant trait but can also follow autosomal recessive or X-linked inheritance.

The Igenomix Hyperekplexia Precision Panel can serve as an accurate and directed diagnostic tool as well as a differential diagnosis of seizures ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Hyperekplexia Precision Panel is indicated in patients with a clinical suspicion or diagnosis presenting with or without the following manifestations:

- Arching of the head alongside the startle reaction
- Jerking movements after startle reaction
- Severe muscle tension
- Lack of movement or slower than normal
- Overactive reflexes
- Intermittent apnea
- Unsteady gait

Clinical Utility

The clinical utility of this panel is:

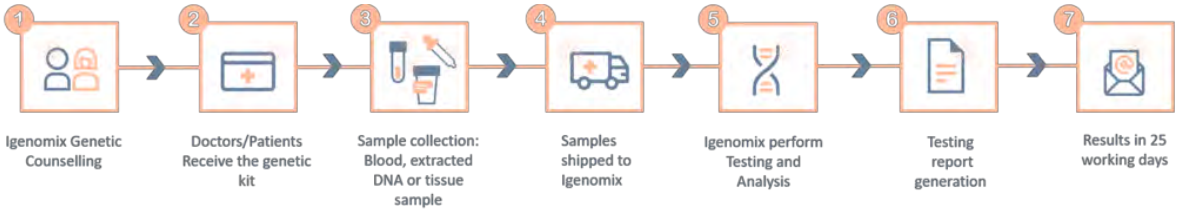
- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of pharmacologic therapy with antiepileptic drugs such as clonazepam.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ARHGEF9	Hyperekplexia, Epilepsy	X,XR,G	100	-
ASNS	Asparagine Synthetase Deficiency	AR	99.98	37 of 37
CACNA1A	Epileptic Encephalopathy, Episodic And Spinocerebellar Ataxia, Migraine, Paroxysmal Torticollis	AD	96.13	249 of 266
CLPB	3-Methylglutaconic Aciduria, Cataracts, Neutropenia	AR	96	26 of 26
CRLF1	Cold-Induced Sweating Syndrome, Crisponi Syndrome, Idiopathic Achalasia	AR	91.53	31 of 33
CTNNB1	Colorectal Cancer, Exudative Vitreoretinopathy, Hepatocellular Carcinoma, Medulloblastoma, Mental Retardation, Pilomatrixoma, Craniopharyngioma, Desmoid Tumor, Spastic Diplegia	AD,AR	100	63 of 63
FKTN	Cardiomyopathy, Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
GLRA1	Hyperekplexia	AD,AR	99.6	71 of 72
GLRB	Hyperekplexia	AR	99.3	16 of 18
GPHN	Hyperekplexia, Molybdenum Cofactor Deficiency	AD,AR	99.2	6 of 6
HEXA	Tay-Sachs Disease	AR	100	205 of 206
RPS6KA3	Coffin-Lowry Syndrome, Mental Retardation	X,XD,G	99.95	-
SCN8A	Cognitive Impairment, Cerebellar Ataxia, Epileptic Encephalopathy, Myoclonus, Seizures, Convulsions, Choreoathetosis	AD	97.85	156 of 172
SLC6A5	Hyperekplexia	AD,AR	100	37 of 37
SLC6A9	Glycine Encephalopathy	AR	99.99	5 of 5
SUOX	Sulfocysteinuria	AR	99.98	28 of 28
TRAK1	Epileptic Encephalopathy	AR	99.28	7 of 7
TSEN54	Encephalopathy, Pontocerebellar Hypoplasia	AR	96.94	20 of 22

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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