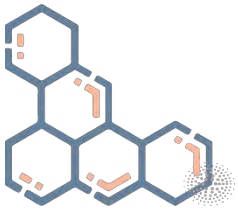
Glycine Encephalopathy

Precision Panel



Overview

Glycine encephalopathy, also known as autosomal recessive nonketotic hyperglycinemia is caused by mutations in the genes encoding components of the glycine cleavage system. It is caused by mutations in the glycine transporter 1 (GLYT1). Glycine encephalopathy an acute metabolic emergency result of an inborn error of metabolism which can result in significant morbidity, progressive neurologic injury, or death. Optimal outcomes of this complication depend upon recognition of the signs and symptoms of metabolic disease, prompt evaluation and referral to a center for evaluation and management. Mutations involved in the glycine cleavage system are inherited in an autosomal recessive pattern.

The Igenomix Glycine Encephalopathy Precision Panel can be used to make an accurate and directed diagnosis 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 Glycine Encephalopathy Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations during the newborn period:

- Respiratory failure requiring mechanical ventilation
- Severe hypotonia progressing to limb hypertonicity
- Startle-like responses
- Dysmorphic features
- Musculoskeletal abnormalities

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment including paediatricians, neurologists, geneticists, genetic counsellors, dietitians, physiotherapists, occupational therapies and orthopaedic surgeons. Dietary restrictions can help prevent acute flares.
- 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**
AMT	Glycine Encephalopathy	AR	99.98	94 of 96
BOLA3	Multiple Mitochondrial Dysfunctions Syndrome	AR	100	8 of 8
GCSH	Glycine Encephalopathy	AR	93.52	1 of 1
GLDC	Glycine Encephalopathy	AR	98.69	359 of 367
GLRX5	Sideroblastic Anemia, Spasticity, Hyperglycinemia	AR	97.17	7 of 8
LIAS	Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	AR	99.82	8 of 8
LIPT1	Lipoyltransferase 1 Deficiency, Leigh Syndrome, Leukodystrophy	AR	97.25	10 of 10
NFU1	Multiple Mitochondrial Dysfunctions Syndrome	AR	100	13 of 15
SLC6A9	Glycine Encephalopathy	AR	99.99	5 of 5

*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

Contact us

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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