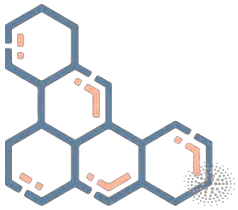
Glycogen Storage Diseases

Precision Panel



Overview

Glycogen Storage Diseases (GSD) are a group of inherited disorders that result from errors of glycogen metabolism caused by mutations in genes that code for enzymes involved in glycogen synthesis and degradation. There are five types of Glycogen Storage Diseases according to their individual enzyme deficiency, type 1 or von Gierke disease being the most common type. The age of onset of symptoms may range from the first months of life to later decades in life. Symptoms arise from abnormal glycogen metabolism and accumulation of glycogen within cells of certain tissues (i.e., liver and skeletal muscles).

The Igenomix Glycogen Storage Diseases Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of hypoglycemia 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 Glycogen Storage Diseases Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Hypoglycemia
- Hepatomegaly
- Poor growth
- Increased uric acid, lactic acid and triglycerides
- Cardiomegaly
- Muscle hypotonia
- Delayed gross motor development
- Respiratory difficulty
- Muscle cramp and fatigue

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

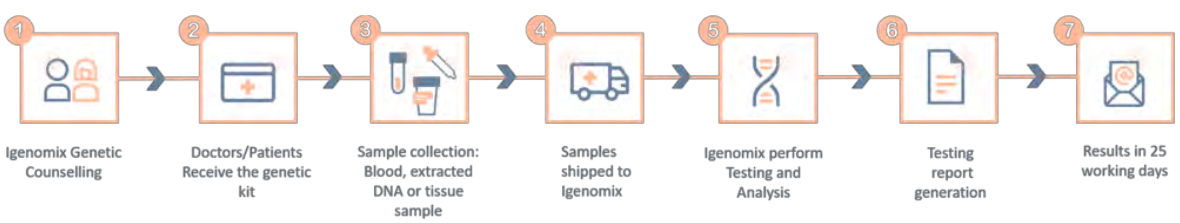
- Early initiation of treatment with a multidisciplinary team in the form of nutritional management with frequent oral glucose consumption and enzyme replacement therapy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AGL	Glycogen Storage Disease Due To Glycogen Debranching Enzyme Deficiency	AR	100	253 of 253
AKT2	Diabetes Mellitus, Hypoinsulinemic Hypoglycemia With Hemihypertrophy, Partial Lipodystrophy	AD	94.99	5 of 6
ALDOA	Glycogen Storage Disease	AR	100	5 of 5
ENO3	Glycogen Storage Disease	AR	100	7 of 7
G6PC	Glycogen Storage Disease	AR	100	125 of 126
GAA	Glycogen Storage Disease	AR	100	623 of 624
GBE1	Glycogen Storage Disease, Polyglucosan Body Disease	AR	99.95	71 of 74
GYG1	Glycogen Storage Disease, Polyglucosan Body Myopathy	AR	100	17 of 18
GYS1	Glycogen Storage Disease	AR	99.69	4 of 4
GYS2	Glycogen Storage Disease	AR	100	24 of 24
HNF4A	Diabetes Mellitus, Fanconi Renotubular Syndrome, Tubulointerstitial Kidney Disease, Hyperinsulinism	AD	100	172 of 174
LAMP2	Danon Disease, Glycogen Storage Disease	X,XD,G	99.96	-
LDHA	Glycogen Storage Disease	AR	99.38	9 of 9
PFKM	Glycogen Storage Disease	AR	99.97	27 of 27
PGK1	Glycogen Storage Disease, Phosphorylase Kinase Deficiency	X,XR,G	100	-
PHKA2	Glycogen Storage Disease, Phosphorylase Kinase Deficiency	X,XR,G	100	-
PHKB	Glycogen Storage Disease	AR	95.99	25 of 25
PHKG2	Glycogen Storage Disease, Phosphorylase Kinase Deficiency	AR	100	33 of 33
PRKAG2	Cardiomyopathy, Glycogen Storage Disease, Wolff-Parkinson-White Syndrome	AD	99.98	61 of 61
PYGL	Glycogen Storage Disease, Glycogen Phosphorylase Deficiency	AR	100	49 of 49
PYGM	Glycogen Storage Disease, Glycogen Phosphorylase Deficiency	AR	100	167 of 169
SLC37A4	Glycogen Storage Disease	AR	99.97	112 of 112

*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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