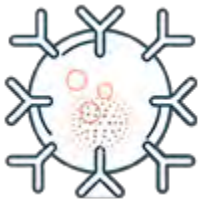
Hemophagocytic Lymphohistiocytosis

Precision Panel



Overview

Hemophagocytic Lymphohistiocytosis (HLH) is a condition where the organism produces too many activated immune cells (macrophages and lymphocytes), creating a state of uncontrolled hyperinflammatory response. Symptoms usually develop within the first months or years of life. Primary HLH are linked to mutations impairing lymphocyte cytotoxicity whereas secondary HLH are triggered by infection, autoimmune disease or neoplasia. Inherited forms of HLH normally involve genes that provide instructions for proteins that help destroy or turn off activated immune cells when they are no longer needed. They are transmitted in an autosomal recessive manner. Early identification of this disease is crucial as it has high rates of morbidity and mortality if under-recognized.

The Igenomix Hemophagocytic Lymphohistiocytosis Precision Panel can be used for an accurate and directed diagnosis as well as differential diagnosis of early recurrent infections 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 Severe Hemophagocytic Lymphohistiocytosis Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Fever
- Splenomegaly
- Neurologic dysfunction
- Coagulopathy
- Liver dysfunction
- Cytopenias
- Hypertriglyceridemia
- Hyperferritinemia

Clinical Utility

The clinical utility of this panel is:

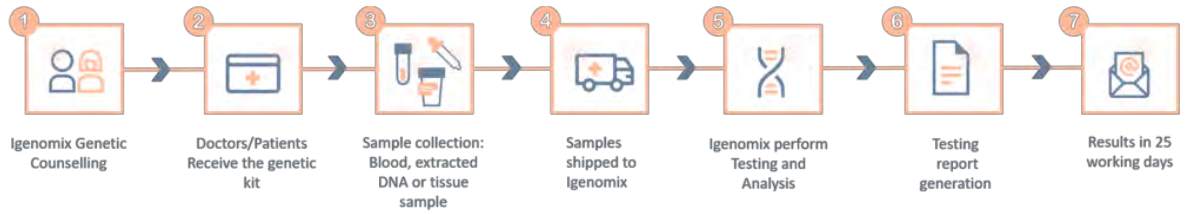
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment involving a multidisciplinary team focusing on haematopoietic stem cell transplantation, medical treatment including immune suppressants, steroids and immunoglobins.
- 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**
AP3B1	Hermansky-Pudlak Syndrome	AR	100	34 of 35
AP3D1	Hermansky-Pudlak Syndrome, Ocular Albinism	AR	99.69	5 of 5
CD27	Lymphoproliferative Syndrome	AR	100	8 of 8
CD70	Lymphoproliferative Syndrome	AR	99.89	4 of 4
CTPS1	Immunodeficiency	AR	100	4 of 4
GATA2	Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Deafness-Lymphedema-Leukemia Syndrome	AD	100	137 of 142
ITK	Lymphoproliferative Syndrome	AR	100	19 of 19
LYST	Chediak-Higashi Syndrome	AR	99.98	117 of 117
MAGT1	Congenital Disorder Of Glycosylation, X-linked Immunodeficiency	X,XR,G	100	-
NLRC4	Autoinflammation With Infantile Enterocolitis, Familial Cold Autoinflammatory Syndrome	AD	99.54	15 of 15
PRF1	Aplastic Anemia, Familial Hemophagocytic Lymphohistiocytosis, Non-Hodgkin Lymphoma, Idiopathic Aplastic Anemia	AR	99.99	196 of 196
RAB27A	Griscelli Syndrome	AR	100	54 of 55
SH2D1A	Lymphoproliferative Syndrome	X,XR,G	99.94	-
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
STX11	Hemophagocytic Lymphohistiocytosis	AR	100	24 of 24
STXBP2	Hemophagocytic Lymphohistiocytosis	AR	99.17	88 of 93
UNC13D	Familial Hemophagocytic Lymphohistiocytosis	AR	99.78	197 of 202
XIAP	X-linked Lymphoproliferative Syndrome	X,XR,G	99.94	-

*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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