Hereditary Hemochromatosis

Precision Panel

Overview

Hereditary hemochromatosis is a condition that results from the body storing an excess of iron. The excess iron is stored in different body organs causing iron overload. This is harmful because the body can only excrete a certain amount of iron, the iron that is not excreted remains in the body and becomes toxic. The organs involved are the liver, heart, pancreas, pituitary, joints, and the skin. There are different types of hemochromatosis that can affect individuals of different ages, all types are inherited in an autosomal recessive manner.

The Igenomix Hereditary Hemochromatosis Precision Panel can be used to make a directed and accurate differential diagnosis of iron overload, 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 Hereditary Hemochromatosis Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of hemochromatosis presenting with the following manifestations:

- Fatigue
- Joint and abdominal pain
- Weight loss
- Lack of sex drive

Clinical Utility

The clinical utility of this panel is:

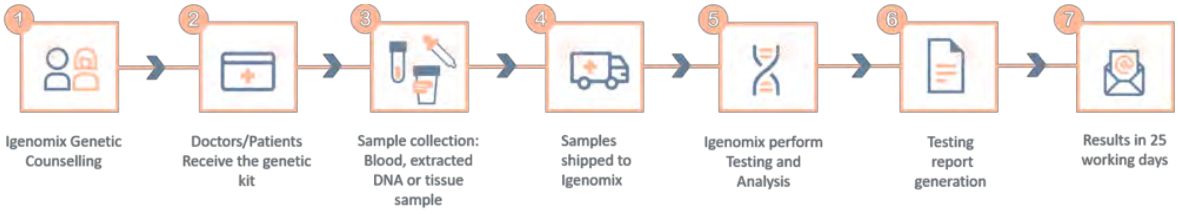
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for early chelation therapy, phlebotomy, surgical intervention and/or dietary considerations and preventive measures.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
BMP2	20p12.3 Microdeletion Syndrome, Brachydactyly Type A2, Hemochromatosis, Short Stature, Facial Dysmorphism and Skeletal Anomalies With or Without Cardiac Anomalies	AD,AR	99.48%	12 of 12
FTH1	Hemochromatosis Type 5	AD	100%	2 of 2
HAMP	Hemochromatosis Type 2B	AR	100%	16 of 17
HFE	Alzheimer Disease, Hemochromatosis Type 1, Pophyria Cutanea Tarda, Porphyria	AD,AR	100%	55 of 57
HJV	Hemochromatosis Type 2A	AR	100%	NA of NA
SLC40A1	Hemochromatosis Type 4	AD	99.80%	62 of 64
TFR2	Hemochromatosis Type 3	AR	99.79%	54 of 54

*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

1. Katsarou, M. S., Papasavva, M., Latsi, R., & Drakoulis, N. (2019). Hemochromatosis: Hereditary hemochromatosis and HFE gene. *Vitamins and hormones*, *110*, 201–222. https://doi.org/10.1016/bs.vh.2019.01.010
2. Pietrangelo A. (2006). Hereditary hemochromatosis. *Biochimica et biophysica acta*, *1763*(7), 700–710. https://doi.org/10.1016/j.bbamcr.2006.05.013
3. Gerhard, G. S., Paynton, B. V., & DiStefano, J. K. (2018). Identification of Genes for Hereditary Hemochromatosis. *Methods in molecular biology (Clifton, N.J.)*, *1706*, 353–365. https://doi.org/10.1007/978-1-4939-7471-9_19