Progressive Familial Intrahepatic Cholestasis

Precision Panel

Overview

Progressive Familial Intrahepatic Cholestasis (PFIC) is an inherited disorder that causes liver damage in the form of cirrhosis and related symptoms due to the accumulation of bile in the liver. There are three types of PFIC that are caused by changes in different genes but have similar symptoms and presentation. Individuals with PFIC1 are more severely affected and have additional health concerns on top of liver disease. Individuals with PFIC2 and PFIC3 show symptoms that are primarily associated with liver disease only and vary in age of onset and severity. All three types of PFIC are inherited in an autosomal recessive manner.

The Igenomix Progressive Familial Intrahepatic Cholestasis Precision Panel can be used to make a directed and accurate differential diagnosis of liver disease, 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 Progressive Familial Intrahepatic Cholestasis Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of hemochromatosis presenting with the following manifestations:

- Itching (pruritus)
- Yellow skin (jaundice)
- Poor growth and weight gains
- Fatigue
- Enlargement of the liver and spleen (hepatomegaly, splenomegaly)

Clinical Utility

The clinical utility of this panel is:

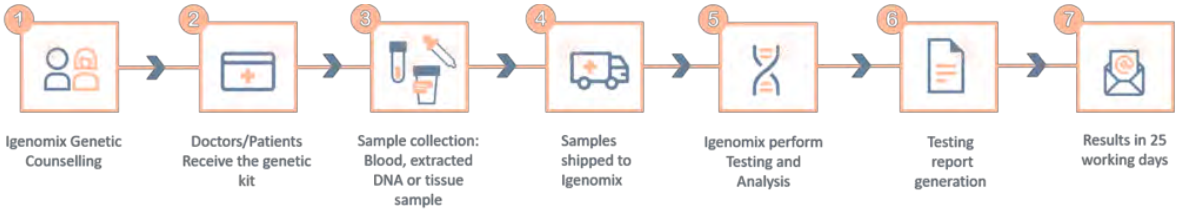
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for early pharmacologic treatment, surgical care, and dietary modifications.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCB11	Benign Recurrent Intrahepatic Cholestasis, Progressive Familial Intrahepatic Cholestasis	AR	99.92%	333 of 334
ABCB4	Intrahepatic Cholestasis Of Pregnancy, Familial Intrahepatic Cholestasis, Gallbladder Disease	AD,AR	100%	255 of 256
ATP8B1	Benign Recurrent Intrahepatic Cholestasis, Intrahepatic Cholestasis Of Pregnancy, Progressive Familial Intrahepatic Cholestasis	AD,AR	99.98%	140 of 140
MYO5B	Diarrhea With Microvillous Atrophy, Microvillus Inclusion Disease	AR	100%	86 of 86
NR1H4	Progressive Familial Intrahepatic Cholestasis	AR	100%	4 of 4
TJP2	Progressive Familial Intrahepatic Cholestasis, Familial Hypercholanemia	AR	99.85%	43 of 43

*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. Bull, L. N., & Thompson, R. J. (2018). Progressive Familial Intrahepatic Cholestasis. *Clinics in liver disease*, *22*(4), 657–669. https://doi.org/10.1016/j.cld.2018.06.003
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