Congenital Hepatic Fibrosis

Precision Panel

Overview

Congenital hepatic fibrosis (CHF) is an inherited or genetic condition that affects the liver and its function. Though CHF can present individually it is often associated with other disorders that can affect kidney function (hepatorenal fibrocystic disease). CHF is caused by malformations and abnormalities in the development of bile ducts starting with the ductal plate. The bile ducts are responsible for ensuring normal blood flow in the vessels of the hepatic portal system as well as the flow of bile. Thus, malformations in this system can disrupt this flow and leads to CHF. This condition is inherited mostly in an autosomal recessive pattern although autosomal dominant and X-linked forms are also found.

The Igenomix Congenital Hepatic Fibrosis Gene Panel can be used to make a directed and accurate differential diagnosis of hepatic fibrosis, ultimately leading to a better management of the comorbidities associated with it and a better 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 Congenital Hepatic Fibrosis Gene Panel is indicated for those patients with a clinical suspicion or diagnosis on hepatic fibrosis with other manifestations including:

- Enlarged liver and spleen (splenomegaly, hepatomegaly)
- Polycystic kidney disease
- Increased blood pressure (hypertension)
- Enlarged kidneys (nephromegaly)
- Inflammation in the bile ducts

Clinical Utility

The clinical utility of this panel is:

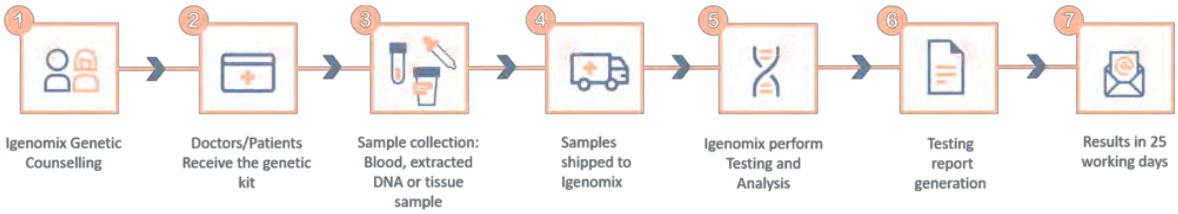
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for early pharmacologic treatment of portal hypertension, surgical care, early referral to specialties like invasive radiology, paediatric nephrology, or vascular surgeon 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**
EFHC1	Juvenile Absence Epilepsy, Juvenile Myoclonic Epilepsy	AD	100%	38 of 39
IL17A	Arthritis, Bonchiolitis Obliterans, Amebiasis, Allergic Contact Dermatitis, Colitis	-	100%	NA of NA
IL17F	Autosomal Dominant Familial Candidiasis, Chronic Mucocutaneous Candidiasis	AD	100%	3 of 3
MCM3	Grade III Astrocytoma, Lung Cancer	-	99.91%	NA of NA
PAQR8	Epilepsy With Generalized Tonic-Clonic Seizures, Myoclonic Juvenile Epilepsy	-	99.98%	1 of 1
PKHD1	Autosomal Recessive Polycystic Kidney Disease	AR	99.97%	582 of 585

*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. Drögemüller, M., Jagannathan, V., Welle, M. M., Graubner, C., Straub, R., Gerber, V., Burger, D., Signer-Hasler, H., Poncet, P. A., Klopfenstein, S., von Niederhäusern, R., Tetens, J., Thaller, G., Rieder, S., Drögemüller, C., & Leeb, T. (2014). Congenital hepatic fibrosis in the Franches-Montagnes horse is associated with the polycystic kidney and hepatic disease 1 (PKHD1) gene. *PloS one*, *9*(10), e110125. https://doi.org/10.1371/journal.pone.0110125
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3. Wen J. (2011). Congenital hepatic fibrosis in autosomal recessive polycystic kidney disease. *Clinical and translational science*, *4*(6), 460–465. https://doi.org/10.1111/j.1752-8062.2011.00306.x