Hyperbilirubinemia

Precision Panel

Overview

The Igenomix Hyperbilirubinemia Precision Panel can be used to make a directed and accurate differential diagnosis of jaundice, 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.

Hyperbilirubinemia is known as high amounts plasma bilirubin levels, causing a yellow discoloration of the skin, sclera, mucous membranes, and other less visible tissues in the newborn. High levels of bilirubin can deposit and accumulate ultimately resulting in neurotoxicity. It can be physiologic or pathologic. There are two types of hyperbilirubinemia depending on the chemical structure of bilirubin: conjugated and unconjugated hyperbilirubinemia. Pathologic congenital causes of hyperbilirubinemia are: Crigler-Najjar syndrome type 1 and 2, Gilbert syndrome, Dubin-Johnson syndrome, and Rotor syndrome. These diseases are inherited mainly in an autosomal recessive pattern.

Indications

The Igenomix Hyperbilirubinemia Precision Panel is indicated for those patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Abdominal distention
- Delayed passage of meconium
- Light-colored stools
- Dark urine
- Infections
- Birth trauma

Clinical Utility

The clinical utility of this panel is:

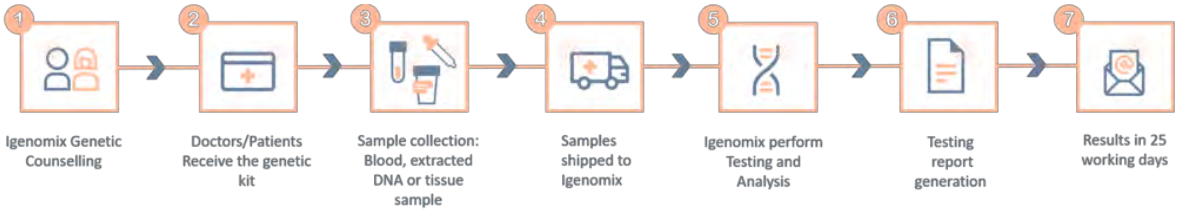
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team for early phototherapy intervention, pharmacologic or even surgical care.
- Risk assessment of asymptomatic family members according to the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

Genes & Diseases

Gene	OMIM Diseases	Inheritance*	% Gene Coverage (20x)	HGMD**
ABCC2	Dubin-Johnson Syndrome	AR	100%	75 of 76
SLCO1B1	Rotor Syndrome	MU,D	99.85%	4 of 4
SLCO1B3	Rotor Syndrome	MU,D	99.30%	4 of 4
UGT1A1	Crigler-Najjar Syndrome Type 1, Crigler-Najjar Syndrome Type 2, Transient Familial Neonatal Breastfeeding Jaundice	AR	na	na

*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. Rets, A., Clayton, A. L., Christensen, R. D., & Agarwal, A. M. (2019). Molecular diagnostic update in hereditary hemolytic anemia and neonatal hyperbilirubinemia. International journal of laboratory hematology, 41 Suppl 1, 95–101. https://doi.org/10.1111/ijlh.13014
2. Watchko J. F. (2013). Genetics and pediatric unconjugated hyperbilirubinemia. *The Journal of pediatrics*, *162*(6), 1092–1094. https://doi.org/10.1016/j.jpeds.2013.01.044