Alagille Syndrome

Gene Sequencing

Overview

Alagille syndrome is a disorder that is associated with problems in several different body organs including – liver disease, congenital heart conditions, abnormalities in the eye and characteristic facial features. Alagille syndrome can present differently, and the clinical symptoms of affected individuals can be very variable. It is characterized by an obstruction of the biliary tree and decreased excretion of bilirubin causing hyperbilirubinemia and jaundice. The different manifestations of Alagille syndrome can be deadly if treatment and management is not administered appropriately. This condition is inherited in an autosomal dominant manner and most cases occur due to a de novo variant. Additionally, Alagille syndrome is associated with variable expression and so the clinical features cannot be predicted with molecular testing.

The Igenomix Alagille Syndrome Gene Sequencing can be used to make a directed and accurate differential diagnosis of jaundice in the newborn, for an early initiation of treatment leading to 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 Alagille Syndrome Gene Sequencing is indicated for those patients with a clinical suspicion or diagnosis of the disease, presenting with the following symptoms:

- Hepatic anomalies
- Renal anomalies
- Cardiac anomalies
- Characteristic facial features
- Eye problems
- Bile duct paucity
- Abnormality in the vertebrae

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE	% GENE COVERAGE (20X)	HGMD*
JAG1	Alagille Syndrome, Tetralogy of Fallot	AD	99.98%	640 of 641
NOTCH2	Acroosteolysis Alagille Syndrome	AD	99.88%	91 of 91

*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

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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