JaundiceChip Resequencing Array

Genes Tested: *ABCB4, ABCB11, ATP8B1, JAG1, SERPINA1*

Inherited intrahepatic cholestasis is a heterogeneous group of disorders typically presenting as neonatal jaundice and leading to persistent liver dysfunction in children and adults. Although some of these conditions are associated with extrahepatic symptoms and varying laboratory and pathologic findings, there remains considerable clinical overlap between these disorders.

Indications:

Diagnostic testing in young patients with intrahepatic cholestasis of unknown etiology. The JaundiceChip resequencing array provides a rapid and cost effective analysis of the most common genetic causes of liver disease in young children.

Specimen:

At least 2 mls of whole blood in purple top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

Methodology:

Combination of a custom designed Affymetrix Resequencing Array and PCR-based confirmational/ rescue sequencing of the coding regions, splice sites and selected untranslated regions of the above specified genes. (PCR-based sequencing of *ABCB4, ABCB11, ATP8B1, JAG1* and *SERPINA1* may also be ordered individually or sequentially—see test requisition).

Clinical Sensitivity:

Disorder	Gene	Estimated Clinical Sensitivity of JaundiceChip*
α-1 Antitrypsin deficiency	*SERPINA1*	>99%
Alagille syndrome	*JAG1*	47%
FIC1 deficiency (PFIC1)	*ATP8B1*	82%
BSEP deficiency (PFIC2)	*ABCB11*	82%
MDR3 deficiency (PFIC3)	*ABCB4*	82%

The clinical sensitivity of the JaundiceChip resequencing array varies by disease. For patients suspected to have Alagille syndrome, we recommend JAG1 PCR-based sequence analysis in lieu of the JaundiceChip resequencing array analysis given the significantly higher detection rate with the PCR-based sequencing test.

*Validation done against 48 patients with known mutations or well-defined clinical diagnosis.

**The sensitivity reported for MDR3 deficiency is based on published reports.

Cincinnati Children's

Human Genetics

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Analytical Sensitivity:

The predicted sensitivity of our JaundiceChip resequencing array is over 99% for the detection of nucleotide base changes or homozygous deletions in the coding exons and exon/intron boundaries of the five genes analyzed. Mutations are confirmed by targeted sequencing of the fragment(s) containing the mutation(s). Heterozygous deletions, insertions, genetic recombinational events, as well as mutations in regulatory regions or other untranslated regions are not detected by this test. If the patient has received a liver transplant or recent blood transfusion, donor DNA may be present in the blood along with patient DNA (chimerism). In this case, additional testing may be required to rule out chimerism.

Note: Single gene sequencing is available for all genes in the panel. Please refer to the test information sheet of these genes on our website for more information.

Turn-Around Times:

- 3-4 weeks

Costs:

Please call 1-866-450-4198 for pricing or with any billing questions.

CPT Codes:

81407, 81479(x4)

Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Shipping Instructions:

Please enclose **test requisition** with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday.

Ship to:

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