HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

Bile Acid Defects Gene Sequencing Panel

This test panel includes 5 genes: ABCD3. AKR1D1, AMACR, CYP7B1, HSD3B7 associated with Congenital bile acid synthesis defect (CBAS) disorders utilizing DNA isolated from a peripheral blood specimen.

Testing Method and Background

This test utilizes **Next Generation Sequencing (NGS) technology,** which provides coverage of all coding exons and noncoding DNA in exon flanking regions (on average 50 bp) enriched using hybrid capture methodology. This assay can detect >99% of described mutations in the included genes, when present, including single nucleotide variants (point mutations), small insertions/deletions (1-25 bp), larger deletions and duplication (<100 bp), complex insertions/deletions, splice site mutations, whole-gene deletions/duplications and exon-level intragenic deletions/insertions in each gene targeted for analysis. All reportable copy number variants are confirmed by independent methodology.

Congenital bile acid synthesis defect (CBAS) disorders are a group of rare inherited metabolic conditions characterized by abnormalities in the production of bile acids. This test aids in the diagnosis of the most common hereditary causes of bile acid synthesis defect disorders, CBAS1-5. The most common symptom of CBAS disorders is cholestasis, where bile flow from the liver is impaired or halted, often accompanied by malabsorption of fat-soluble vitamins.

Highlights of Bile Acid Defects Gene Sequencing Panel

Targeted Region

ABCD3, AKR1D1, AMACR, CYP7B1, HSD3B7

- **Wide-ranging Coverage of Variants**
  Detects and provides coverage of all coding exons and noncoding DNA in exon flanking regions.
- **Accurate Results Using Clinically Validated Computational Data Analysis**
  A variety of mutation types (point, indels and duplications) are confirmed using computational data analysis for sequence variant calling, filtering and annotation.

Ordering Information

Get started (non-HFHS): Print a Hereditary Cancer Panels requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "Bile Acid Defects Gene Sequencing Panel" (DNA2100035)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Extracted DNA - from a CLIA-certified Laboratory

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 10-14 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81479, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

Revision: 1; 12-04-2024