HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

Cystic Diseases of Liver/Kidney Gene Sequencing Panel

This test panel includes 10 genes: ALG5, ALG8, ALG9, IFT140, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63 associated with Polycystic liver disease 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.

Polycystic liver disease is a collection of rare human disorders that result from structural changes in the biliary tree development characterized by bilateral kidney cysts, liver cysts, and an increased risk of intracranial aneurysms. This test aids in the diagnosis of the most common hereditary causes of polycystic kidney disease, PKD1. Autosomal dominant polycystic liver disease (PCLD) is caused by heterozygous pathogenic variants in PRKCSH, SEC63, ALG8 or LRP5 gene. Similarly, polycystic kidney disease is a collection of rare human disorders characterized by bilateral kidney cysts, liver cysts, and an increased risk of intracranial aneurysms. Autosomal dominant polycystic kidney disease (ADPKD) is caused by heterozygous pathogenic variants in PKD1 and/or PKD2 in majority of cases. More rarely, ADPKD is caused by a heterozygous pathogenic variant in ALG5, ALG9, or IFT140 and several other rare genetic causes. Autosomal recessive polycystic kidney disease is caused by biallelic mutations in PKHD1 and is characterized by primary involvement of the kidneys and liver with mostly secondary effects seen in other organ systems.

Highlights of Cystic Diseases of Liver/Kidney Gene Sequencing Panel

Targeted Region

ALG5, ALG8, ALG9, IFT140, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63

- **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 Germline/Non-Cancer Hereditary Test requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "Cystic Diseases of Liver/Kidney Gene Sequencing Panel" (DNA2100036)

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

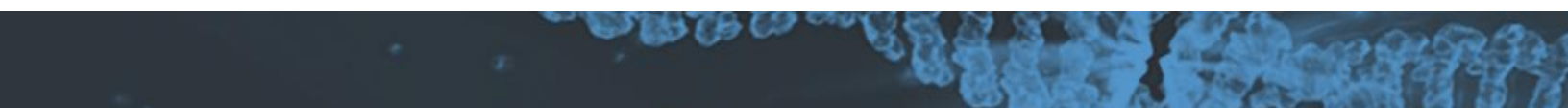
Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

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: 81407, 81406, 81408, 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

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