HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

IDH1 Mutation Detection (Hematolymphoid)

IDH1 Mutation Detection provides targeted detection of mutations in the IDH1 gene using DNA extracted from blood or bone marrow specimens. This test is used for diagnostic, prognostic, and predictive purposes associated with Hematological disorders.

Testing Method and Background

This test utilizes hybrid capture method followed by **Next Generation Sequencing (NGS) technology** for detection of mutations in isocitrate dehydrogenase (NADP(+)) 1 (IDH1) gene associated with Hematologic malignancies (tumor suppressor genes and oncogenic hot spots) using extracted DNA from blood or bone marrow specimens. Data analysis provides variant detection and annotation, interpretation of clinically significant genomic alterations and their association to approved or investigational therapies. This assay is designed to detect single nucleotide variants, insertions, deletions and copy number alterations within the defined target regions. Variants outside the define regions may not be detected.

Acute myeloid leukemia (AML) is a clinically and genetically heterogeneous disease that has poor prognosis. Combinations of mutations interact to drive the initiation and progresion of AML and may create unique sensitivities to epigenetic-focused and other targeted or chemotherapies. Approximately 12% of AML patients have mutations in IDH2. IDH1 mutations are slightly less common. The phenotype of IDH1 and IDH2 mutant AML is similar, characterized by the gain-of-function activity, which impairs cellular differentiation.

Highlights of IDH1 Mutation Analysis (Hematolymphoid)

Targeted Region

IDH1: Exons 3-10

- **Accurate Results from Low-Quality Samples**
  Workflow with low quantity of input DNA and accurate detection of variants down to 5% mutant allele frequency.
- **Wide-ranging Coverage of Variants**
  Assessment of single-nucleotide variants (SNVs) and small insertions/deletions within multiple target exons

Ordering Information

Get started (non-HFHS): Print a Molecular Hematologic Testing requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "IDH1 Mutation Detection" (MOL80511 Blood, MOL80512 Bone Marrow)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Bone Marrow - 1-3ml, anticoagulated with either heparin or EDTA, **Specimen stability: Refrigerated - 1 week** (ship cold)
- 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: 5-10 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: 81120, 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: 2; 11-27-2024