HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

Solid Tumor Mutation IDH1

This test detects mutations in the IDH1 gene from DNA extracted from formalin-fixed, paraffin-embedded (FFPE) specimens. This test is used for diagnostic, prognostic, and predictive purposes associated with Glioma tumors.

Testing Method and Background

The gene target exons are enriched by hybrid capture method followed by Next Generation Sequencing (NGS). This method was optimized for use with low quantity of input DNA (50 ng) obtained from formalin-fixed, paraffin-embedded (FFPE) tissues providing high on-target coverage with coverage uniformity above 95% throughout the entire target region.

Brain tumors (gliomas) with IDH1 or IDH2 mutations have distinctive genetic and clinical characteristics, and patients with such tumors have a better outcome than those with wild type IDH genes. Mutation of IDH1 occurs early in glioma progression with somatic mutations of the R132 residue of IDH1 identified in majority (>70%) of grades II and III astrocytomas and oligodendrogliomas, as well as in secondary GBMs that developed from these lower grade lesions. Mutation analysis of closely related IDH2 revealed mutations of IDH2 residue R172 or R140, with most mutations occurring in tumors lacking IDH1 mutations.

Highlights of Solid Tumor Mutation IDH1

Targeted Region

IDH1: Exon 4

- **Accurate Results from Low-Quality Samples**
  Sensitive variant detection with as little as 50 ng of input DNA, and as low as 5% mutant allele frequency, maximizes the results from low input sample types such as formalin fixed, paraffin embedded (FFPE) sections.
- **Wide-ranging Coverage of Variants**
  Assessment of single-nucleotide variants (SNVs) and small insertions/deletions, and whole gene deletions and amplifications.

Ordering Information

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

Get started (HFHS): Order through Epic using test "IDH1 Mutation" (MOL8011)

Specimen requirements:

A surgical pathologist should confirm the presence of adequate tumor in materials submitted for analysis. Section from archival paraffin material or frozen surgical biopsies should be confirmed to contain >50% tumor by a surgical pathologist. If the submitted material for analysis contains < 50% of tumor, areas of predominant tumor will be microdissected, if possible, to enrich for neoplastic cells.

- Formalin-fixed, paraffin-embedded tissue, preferably no older than 2 years
- 5-6 tissue sections at 5-6 micron thickness (please include H&E slide and a copy of pathology report)
- Cytology slides (cell block with 500+ tumor cells, submit block or 5-6 tissue sections at 5-10 micron thickness depending on cellularity)
- Extracted DNA - from a CLIA-certified Laboratory

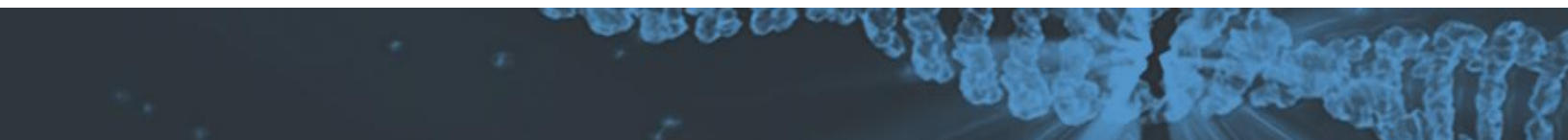
Cause for Rejection: Fresh unfixed tissue, paraffin materials that do not contain tumor cells, improperly labeled specimens, archival paraffrin material subjected to acid decalcification.

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

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