IDH1/IDH2 Mutation Analysis

IDH1 Result
NEGATIVE
No IDH1 mutations were detected in the provided specimen. This result does not rule out the presence of IDH mutations at a level below the detection sensitivity of the assay, or the presence of other mutations within IDH not detected by this assay. Results should be interpreted in conjunction with clinical and other laboratory findings for the most accurate interpretation.

IDH2 Result
NEGATIVE
No IDH2 mutations were detected in the provided specimen. This result does not rule out the presence of IDH mutations at a level below the detection sensitivity of the assay, or the presence of other mutations within IDH not detected by this assay. Results should be interpreted in conjunction with clinical and other laboratory findings for the most accurate interpretation.

Indication
Comment: 01
BRAIN, GLIOMA

Location
Comment: 01
LEFT FRONTAL BRAIN

Specimen Type
Formalin-fixed, paraffin-embedded tissue block

Block ID
Comment: 01
TEST123

Background
Isocitrate dehydrogenase 1 and 2 (IDH1 and IDH2) are the most frequently mutated metabolic genes in human cancer. They encode cytosolic and mitochondrial enzymes that catalyze the conversion of isocitrate to α-ketoglutarate (αKG), a key component in metabolic and cellular pathways including the Krebs cycle. IDH1 and IDH2 mutations are found in multiple types of human cancer including, but not limited to, acute myeloid leukemia and gliomas. Identification of IDH mutations can aid in their diagnosis, provide prognostic information, and suggest treatment with IDH inhibitors. This assay will detect mutations affecting amino acids 100, 105, and 132 of IDH1, and amino acids 140 and 172 of IDH2.

Method
Genomic DNA was isolated from the provided tumor specimen. The
TESTS

RESULTS

FLAG

UNITS

REFERENCE INTERVAL LAB

IDH1 and IDH2 gene region of interest were subjected to SNaPshot multiplex PCR and primer extension for mutation detection. This assay is able to detect 5% mutation in a background of wild-type DNA.

This test can detect the following mutations in IDH1 and IDH2


References


(6) https://www.fda.gov/Drugs/InformationonDrugs/ApprovedDrugs/ucm569482.htm

(7) https://www.fda.gov/Drugs/InformationOnDrugs/ApprovedDrugs/hematologyoncology-cancer-approvals-safety-notifications

Director Review

Dan Wang, PhD, FACMG
Director, Molecular Oncology
LabCorp Center for Molecular Biology and Pathology
Research Triangle Park, NC 27709
1-800-533-0567

Disclaimer:
This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration.

Microdissection Performed
Completed

For inquiries, the physician may contact Branch: 800-762-4344 Lab: 800-735-4087
A duplicate report has been generated due to demographic updates.

**Ordered Items**  
IDH1/IDH2 Mutation Analysis

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<tr>
<td>IDH1 Result</td>
<td>POSITIVE</td>
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<tr>
<td>An IDH mutation was detected in the provided specimen. The frequency of IDH mutations in acute myeloid leukemia is 19% and 7% in myelodysplastic syndrome (MDS). IDH mutations are associated with worse prognosis in MDS and myeloproliferative neoplasms. The FDA has approved an oral targeted inhibitor of IDH2, enasidenib (IDHIFA(R)), for the treatment of adult patients with relapsed or refractory AML (R/R AML) with IDH2 mutations. The FDA has approved an oral targeted inhibitor of IDH1, ivosidenib (TIBSOVO(R)), for the treatment of adult patients with relapsed or refractory AML (R/R AML) with IDH1 mutations. The FDA approved ivosidenib (TIBSOVO(R)), for newly-diagnosed acute myeloid leukemia (AML) with a susceptible IDH1 mutation in patients who are at least 75 years old or who have comorbidities that preclude the use of intensive induction chemotherapy. Results should be interpreted in conjunction with clinical and other laboratory findings for the most accurate interpretation.</td>
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<td>IDH1 Nuc Change</td>
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<td>IDH1 Amino Acid Change</td>
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Results should be interpreted in conjunction with clinical and other laboratory findings for the most accurate interpretation.

**IDH2 Nuc Change**
c.419G>A 01

**IDH2 Amino Acid Change**
p.R140Q 01

**Indication**
ACUTE MYELOID LEUKEMIA

**Location**
PERIPHERAL BLOOD

**Specimen Type**
Bone Marrow.

**Block ID**
N/A

**Background**
Isocitrate dehydrogenase 1 and 2 (IDH1 and IDH2) are the most frequently mutated metabolic genes in human cancer. They encode cytosolic and mitochondrial enzymes that catalyze the conversion of isocitrate to a-ketoglutarate (alphaKG), a key component in metabolic and cellular pathways including the Krebs cycle. IDH1 and IDH2 mutations are found in multiple types of human cancer including, but not limited to, acute myeloid leukemia and gliomas. Identification of IDH mutations can aid in their diagnosis, provide prognostic information, and suggest treatment with IDH inhibitors. This assay will detect mutations affecting amino acids 100, 105, and 132 of IDH1, and amino acids 140 and 172 of IDH2.

**Method**
Genomic DNA was isolated from the provided tumor specimen. The IDH1 and IDH2 gene region of interest were subjected to SNaPshot multiplex PCR and primer extension for mutation detection. This assay is able to detect 5% mutation in a background of wild-type DNA.

This test can detect the following mutations in IDH1 and IDH2


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<td>Reardon D, Herndon J, Kinzler KW, Velculescu VE, Vogelstein B, Bigner DD.</td>
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<th>01 YU LabCorp RTP</th>
<th>Dir: Arundhati Chatterjee, MD</th>
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<td>1904 TW Alexander Drive Ste C, RTP, NC 27709-0153</td>
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