

Specimen Collected: 14-Sep-21 14:07

IDH1 and IDH2 Mutation Analysis | Received: 14-Sep-21 14:08

Report/Verified: 14-Sep-21 14:13

FFPE

Procedure	Result	Units	Reference Interval
IDH1 and IDH2 Mutation	Detected ^{f1 i1}		
Results			
IDH1-2 FFPE Source	FFPE Tissue		
Block ID	BCDE		

Result Footnote

f1: IDH1 and IDH2 Mutation Results

A mutation in IDH2 exon 4 was detected: p.R172K

This result has been reviewed and approved by Anna Matynia, M.D.

Test Information

i1: IDH1 and IDH2 Mutation Results

BACKGROUND INFORMATION: IDH1 and IDH2 Mutation Results

CHARACTERISTICS: This test is designed to detect mutations in exon 4 of the IDH1 and IDH2 genes at "hotspots" R132 of IDH1 and R140 and R172 of IDH2 that are frequently present in gliomas and in a subset of cases of acute myeloid leukemia. IDH1/2 mutations in gliomas are generally associated with a better prognosis. In acute myeloid leukemia, the prognostic significance of IDH1 mutations is context dependent. IDH1 mutations appear to be associated with worse outcome in patients without FLT3-ITD mutations (see J Clin Oncol 2010. 28:3636 and Blood 2010. 116:2779). In acute myeloid leukemia patients with IDH2 abnormalities, IDH2 R140 mutations appear to be associated with better outcome while IDH2 R172 mutations appear associated with worse outcome (see Blood 2011. 118:409). The FDA has approved ivosidenib as a targeted therapy for acute myeloid leukemia with an IDH1 mutation, and enasidenib for AML with an IDH2 mutation. Clinical trials may be available.

METHODOLOGY: DNA is isolated from FFPE tissue, blood, or bone marrow. The DNA is amplified for IDH1 and IDH2 covering exon 4 of both genes including the important residues R132 (IDH1), R140 (IDH2) and R172 (IDH2). Sanger sequencing is then performed to detect mutations. Only mutations in R132 (IDH1), R140 and R172 (IDH2) are reported.

LIMITATIONS: Mutations in other locations within the IDH1 and IDH2 genes or in other genes will not be detected. The limit of detection for this test is 20 percent mutant allele. Results of this test must always be interpreted within the clinical context and with other relevant data, and should not be used alone for a diagnosis of malignancy. This test is not intended to detect minimal residual disease.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H=High, i=Test Information, L=Low, t=Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Tracy I. George, MD

ARUP Accession: 21-257-900245**Report Request ID:** 15047860**Printed:** 14-Sep-21 14:14

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i1: IDH1 and IDH2 Mutation Results

Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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