
Specimen Collected: 12-Mar-24 15:31

IDH1 and IDH2 Mutation Detection Procedure	Received: 12-Mar-24 15:31	Report/Verified: 18-Mar-24 17:48
Procedure	Result	Reference Interval

IDH1-IDH2 Int	Detected ^{f1 i1}	
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IDH1 and IDH2 Mutation Detection Procedure	Received: 12-Mar-24 15:31	Report/Verified: 19-Mar-24 06:38
Procedure	Result	Reference Interval

Block ID	1	
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Result Footnote

f1: IDH1-IDH2 Int

IDH1 and IDH2 Mutation Detection

A mutation in IDH1 was detected: c.395G>A, p.Arg132His

This result has been reviewed and approved by Rakhi Jattani, Sequencing Analyst.

Test Information

i1: IDH1-IDH2 Int

BACKGROUND INFORMATION: IDH1 and IDH2 Mutation Detection

CHARACTERISTICS: This assay is an amplicon enrichment-based massively parallel sequencing assay targeting hotspot variants in genes critical for the diagnostic, prognostic, and therapeutic assessment of various solid tumors. The amplicon primer pool is designed to interrogate variants within a limited set of highly clinically relevant gene loci for the identification of actionable somatic variants in FFPE tissue from solid tumors.

GENES TESTED: IDH1 (NM_005896) exon 4 and IDH2 (NM_002168) exon 4 are evaluated to detect hotspot variants. Targeted regions include chr2:209113083-209113124, chr15:90631809-90631869, and chr15:90631901-90631989.

METHODOLOGY: Genomic DNA was isolated from a microscopically-guided dissection of FFPE tumor tissue and then enriched for the targeted regions of the tested genes. The variant status of the targeted genes was determined by massively parallel sequencing. The hg19 (GRCh37) reference sequence was used as a reference for identifying genetic variants. Clinically significant single nucleotide variants and variants of uncertain significance within the preferred transcripts are reported. Other types of variants may be reported with a disclaimer, if detected.

LIMITATIONS: This test will not detect variants in areas outside the targeted genomic regions or below the limit of detection. More information about the targeted regions of this test is included in the Additional Technical Information available in the Laboratory Test Directory. Copy number alterations (losses or amplifications), translocations, microsatellite instability, tumor mutational burden, deep intronic variants, and insertions/deletions will not be detected. Since this is a DNA-based assay, RNA variants will not be detected. This test evaluates

*=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: Jonathan R. Genzen, MD, PhD

ARUP Accession: 24-072-900222**Report Request ID:** 19133907**Printed:** 21-Mar-24 15:07

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Test Information

i1: IDH1-IDH2 Int

for variants in tumor tissue only and cannot distinguish between somatic and germline variants. Therefore, if a hereditary/familial cancer is of clinical concern, additional clinical evaluation and genetic counseling should be considered prior to additional testing. In some cases, variants may not be identified due to technical limitations related to the presence of known pseudogenes, GC-rich regions, repetitive or homologous regions, low mappability regions, and/or variants located in regions overlapping amplicon primers. Tissue samples yielding between 1ng and 5ng total DNA input may yield suboptimal results and will be accepted for testing with a client-approved disclaimer. Benign or likely benign variants in the preferred transcript are not reported. Variant allele frequency (VAF) is not reported. Additional evaluation should be considered for complete genetic analysis, including detection of variants outside of the hotspot regions of IDH1 or IDH2, variants within other genes, gene methylation, translocations, or gene rearrangements, if clinically indicated.

LIMIT OF DETECTION (LOD): The LOD for this assay is 10 percent VAF. For variants near the assay LOD, positive percent agreement (PPA) was found to be greater than 90 percent.

ANALYTICAL ACCURACY/SENSITIVITY (PPA): The PPA estimate for the relevant variant class (with 95 percent credibility region) is listed below. Genes included on this test are a subset of a larger methods-based validation from which the PPA values are derived.

Single nucleotide variants (SNVs): 98.4 percent (95.1-99.7 percent)

CLINICAL DISCLAIMER: Results of this test must always be interpreted within the context of clinical findings and other relevant data and should not be used alone for a diagnosis of malignancy, determination of prognosis, or recommendation of therapy. 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 U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

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