

BRAF and KIT Mutation Analysis, Next-Generation Sequencing, Tumor

Patient ID SA00174967	Patient Name TESTINGRNV, ABNORMAL	Birth Date 1985-01-05	Sex F	Age 40
Order Number SA00174967	Client Order Number SA00174967	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 28 Mar 2025 00:00		

BRAF/KIT Mutation Analysis, Tumor

Result

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Provided diagnosis: melanoma involving soft tissue

The following CLINICALLY RELEVANT VARIANT was detected:

Gene: BRAF

DNA Change: c.1799T>A (Exon 15)

Amino Acid Change: p.V600E (Val600Glu)

Variant Allele Frequency: 15.8%

No other reportable sequence variants were detected within the analyzed regions of the tested gene(s) listed in the method description.

Interpretation

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BRAF c.1799T>A (p.V600E) (Exon 15)

BRAF encodes the signaling protein Braf, which is downstream of Ras and activates the MAPK pathway. Braf signaling is critically involved in the processes of cell division and differentiation. BRAF activating mutations occur predominantly at a single location (V600E). BRAF activating mutations or amplification have been reported to result in uncontrolled cell growth and tumorigenesis [PMID:12068308, PMID:21447722].

Clinically approved targeted therapy is available for patients with unresectable or metastatic melanoma with BRAF V600 mutations [fda.gov/drugs].

Additional Information

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CLINICAL TRIALS

Possible clinical trials of benefit for this patient can be found at the following sites:

1) ClinicalTrials.gov:

www.clinicaltrials.gov/ct2/search/advanced

2) Mayo Clinic: www.mayo.edu/research/clinical-trials/ 3) National Cancer Institute: www.cancer.gov/clinicaltrials/search

REFERENCE TRANSCRIPT

Sequence variant nomenclature is based on the following RefSeq accession number (build GRCh37 (hg19)): BRAF NM_004333.

Specimen

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Tissue, Tumor

Tissue ID

MCR

13245

Method

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Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection. DNA is extracted from FFPE or cytology slides, and next generation sequencing is performed to evaluate the presence of a mutation in all coding regions and exon/intron boundaries of the BRAF and KIT genes.

Variant nomenclature is based on build GRCh37 (hg19). For details about gene transcripts (RefSeq accession numbers), specific targeted regions of each gene, and additional information on this test, see www.mayocliniclabs.com (Test ID BRFKT).

Disclaimer

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This test cannot differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292



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DNA variants of uncertain significance may be identified.

A negative result does not rule out the presence of a variant that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for sequence reportable alterations is 5% mutant allele frequency with a minimum coverage of 500X in a sample with at least 20% tumor content.

Point mutations and small insertion/deletion mutations will be detected in the BRAF and KIT genes only. This test may detect single exon deletions but does not detect multi-exon deletions, duplications, larger-scale genomic copy number variants, copy neutral loss of heterozygosity (LOH), or epigenetic modifications such as promoter methylation. DELINS of 1,000 bp or less are detectable with at least 50 or more supporting reads.

Variant allele frequency (VAF) is the percentage of sequencing reads supporting a specific variant divided by the total sequencing reads at that position. In somatic testing, VAF should be interpreted in the context of several factors including, but not limited to: tumor purity/heterogeneity/copy number status (ploidy, gains/losses, loss of heterozygosity) and sequencing artifact/misalignment [PMID: 27144058, PMID: 29769535].

Rare polymorphisms may be present that could lead to false-negative or false-positive results.

The presence or absence of a variant may not be predictive of response to therapy in all patients.

Test results should be interpreted in the context of clinical, tumor sampling, histopathological, and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for discussion. Misinterpretation of results may occur if the information provided is inaccurate and/or incomplete.

Reliable results are dependent on adequate specimen collection and processing. This test has been validated on cytology slides and formalin-fixed, paraffin-embedded tissues; other types of fixatives are discouraged. Improper treatment of tissues, such as decalcification, may cause PCR failure.

Released By

Sounak Gupta, M.B.B.S., Ph.D.

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Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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