MAYO CLINIC LABORATORIES

1-800-533-1710

CTNBT

B-Catenin Mutation Analysis, Next-Generation Sequencing, Tumor

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174969	TESTINGRNV, ABNORMAL		1986-01-06	F	39
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174969	SA00174969	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

B-Catenin Mutations Analysis, Tumor

Result

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Provided diagnosis: soft tissue bland myofibroblastic proliferation

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: CTNNB1
DNA Change: c.134C>T (Exon 3)
Amino Acid Change: p.S45F (Ser45Phe)
Variant Allele Frequency: 32%

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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CTNNB1 c.134C>T (p.S45F) (Exon 3)

CTNNB1 encodes the beta-catenin protein. CTNNB1 mutations, particularly those in exon 3 (amino acids 5–81; Integrative Genomics Viewer, v.2.8), have been associated with increased beta-catenin protein stability and activation of the Wnt pathway [PMID:10072352, PMID:11943721, PMID:11731417, PMID:16523258, PMID:20126258, PMID:15060161]. CTNNB1 can act as an oncogene and altered expression of beta-catenin can lead to abnormal signaling in various diseases, including modulation of gene transcription to drive cancer initiation, progression, survival, and relapse [PMID:23490077].

Mutations in CTNNB1 are seen in multiple tumor types, including: desmoid-type fibromatosis, beta-catenin activated hepatocellular adenoma, hepatocellular carcinoma, solid pseudopapillary neoplasms of the pancreas, Sertoli cell tumors (sex cord stromal tumors), medulloblastoma, and adamantinomatous craniopharyngioma [COSMIC, cBioPortal for Cancer Genomics].

The PRESENCE of a mutation in CTNNB1 supports the diagnosis of any of these tumor types and should be interpreted in correlation with histopathological and clinico-radiological findings and other genetic test results.

There are currently no known clinically approved therapies that specifically target CTNNB1 mutations.

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)):CTNNB1 NM_001904.

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 BCAT (CTNNB1) gene.

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 CTNBT).

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

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174969	TESTINGRNV, ABNORMAL		1986-01-06	F	39
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Disclaimer

[1] MCR

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.

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 BCAT (CTNNB1) gene 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

MCR

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

Received: 29 Mar 2025 10:06 **Reported:** 06 Apr 2025 11:30

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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