Overview

Useful For
Distinguishing desmoid-type fibromatosis from other soft tissue tumors by assessing gene targets with in the BCAT (CTNNB1) gene.

This test is not useful for hematological malignancies.

Additional Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>SLIRV</td>
<td>Slide Review in MG</td>
<td>No, (Bill Only)</td>
<td>Yes</td>
</tr>
</tbody>
</table>

Testing Algorithm

When this test is ordered, slide review will always be performed at an additional charge.

Special Instructions

- Tissue Requirements for Solid Tumor Next-Generation Sequencing

Method Name
Polymerase Chain Reaction (PCR)-Based Next-Generation Sequencing

NY State Available
Yes

Specimen

Specimen Type
Varies

Necessary Information
Pathology report (final or preliminary) at minimum containing the following information must accompany specimen in order for testing to be performed:

1. Patient name
2. Block number—must be on all blocks, slides and paperwork (can be handwritten on the paperwork)
3. Tissue collection date
4. Source of the tissue

Specimen Required
This assay requires at least 20% tumor nuclei.

- Preferred amount of tumor area with sufficient percent tumor nuclei: tissue 144 mm(2)
- Minimum amount of tumor area: tissue 36 mm(2).
- These amounts are cumulative over up to 10 unstained slides and must have adequate percent tumor nuclei.
- Tissue fixation: 10% neutral buffered formalin, not decalcified
- For specimen preparation guidance, see Tissue Requirement for Solid Tumor Next-Generation Sequencing in Special Instructions. In this document, the sizes are given as 4mm x 4mm x 10 slides as preferred: approximate/equivalent to 144 mm(2) and the minimum as 3mm x 1mm x 10 slides: approximate/equivalent to 36mm(2).

Preferred:

**Specimen Type:** Tissue block  
**Collection Instructions:** Submit a formalin-fixed, paraffin-embedded tissue block with acceptable amount of tumor tissue.

Acceptable:

**Specimen Type:** Tissue slide  
**Slides:** 1 stained and 10 unstained  
**Collection Instructions:** Submit 1 slide stained with hematoxylin and eosin and 10 unstained, nonbaked slides with 5-micron thick sections of the tumor tissue.

Note: The total amount of required tumor nuclei can be obtained by scraping up to 10 slides from the same block.

**Specimen Type:** Cytology slide (direct smears or ThinPrep)  
**Slides:** 1 to 3 slides  
**Collection Instructions:** Submit 1 to 3 slides stained and cover slipped with a preferred total of 5000 nucleated cells or a minimum of at least 3000 nucleated cells.

Note: Glass coverslips are preferred; plastic coverslips are acceptable but will result in longer turnaround times.

**Additional Information:** Cytology slides will not be returned.

**Forms**
If not ordering electronically, complete, print, and send an Oncology Test Request (T729) with the specimen.

**Specimen Minimum Volume**
See Specimen Required

**Reject Due To**
Test Definition: CTNNB
B-Catenin Mutation Analysis, Tumor

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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</thead>
<tbody>
<tr>
<td>Varies</td>
<td>Ambient (preferred)</td>
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<td></td>
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<tr>
<td>Frozen</td>
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<tr>
<td>Refrigerated</td>
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Specimen Stability Information

Clinical and Interpretive

Clinical Information
Desmoid-type fibromatosis is a locally invasive soft tissue tumor. The histological diagnosis of desmoid-type fibromatosis is challenging. Mutations in exon 3 of the beta-catenin (BCAT also known as CTNNB1) gene have been identified in 50% to 87% of desmoid-type fibromatosis, including T41A (121 A->G), S45P (133 T->C), and S45F (134 C->T), but not in other soft tissue tumors. Patients harboring beta-catenin mutations may have a higher recurrence rate compared to the patients with wild-type beta-catenin.

Next-generation sequencing has recently emerged as an accurate, cost-effective method to identify alterations across numerous genes. This test uses formalin-fixed paraffin-embedded tissue or cytology slides to assess for common somatic mutations in the beta-catenin gene known to be associated with desmoid-type fibromatosis. The results of this test can be useful for supporting a diagnosis of desmoid-type fibromatosis and predicting prognosis.

Reference Values
An interpretative report will be provided.

Interpretation
An interpretative report will be provided.

Cautions
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 (wild-type) result does not rule out the presence of a mutation that may be present but below the limits of detection of this assay.

Point mutations and small insertion/deletion mutations will be detected with in the BCAT (CTNNB1) gene only. This test does not detect large single or multiexon deletions, or duplications or genomic copy number variants.

Rare polymorphisms may be present that could lead to false-negative or false-positive results. Test results should be interpreted in the context of clinical findings, tumor sampling and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for updated interpretation. Misinterpretation of results may occur if the information provided is inaccurate 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.

**Supportive Data**

This next-generation sequencing assay detects somatic mutations that can be used to assist in the diagnosis of desmoid-type fibromatosis and predicting prognosis.

This assay has been shown to be very reproducible, having a 100% concordance for intra- and interassay reproducibility experiments. All somatic mutations that had been previously identified by various other molecular methods were detected by this assay during accuracy studies. No pathogenic variants were detected in known mutation-negative samples.

**Clinical Reference**


**Performance**

**Method Description**

Next-generation sequencing is performed to test for the presence of a mutation in targeted regions of the BCAT (CTNNB1) gene, including exon 3, codons 25-66. (Unpublished Mayo method).

<table>
<thead>
<tr>
<th>Gene</th>
<th>GenBank Accession Number</th>
<th>Nucleotide Start</th>
<th>Nucleotide End</th>
<th>Chromosome</th>
<th>Exon</th>
<th>Codons</th>
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<td>NM_001904</td>
<td>41266076</td>
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<td>Chromosome 3</td>
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**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Monday through Friday; Varies

**Analytic Time**

12 days

**Maximum Laboratory Time**

20 days
Specimen Retention Time
Unused portions of blocks will be returned to the client. Unused slides are stored indefinitely.

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
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.

CPT Code Information
81403
88381

LOINC® Information

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<td>B-Catenin Mutation Analysis, Tumor</td>
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