Overview

Useful For
Aiding in the diagnosis of myxoid/round cell liposarcoma by detecting a neoplastic clone associated with gene rearrangement involving the DDIT3 (CHOP) gene region at 12q13

Reflex 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>_PBCT</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PADT</td>
<td>Probe, +1</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PB02</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PB03</td>
<td>Probe, +3</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_IL25</td>
<td>Interphases,</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_I099</td>
<td>Interphases, 25-99</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_I300</td>
<td>Interphases, &gt;=100</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
</tbody>
</table>

Testing Algorithm
This test includes a charge for application of the first probe set (2 FISH probes) and professional interpretation of results. Additional charges will be incurred for application of all reflex probes performed. Analysis charges will be incurred based on the number of cells analyzed per probe set. If no cells are available for analysis, no analysis charges will be incurred.

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Tissue

Advisory Information
This test does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered and the appropriate FISH test will be ordered and performed at an additional charge.

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information
A reason for referral and pathology report are required in order for testing to be performed. Send information with specimen. Acceptable pathology reports include working drafts, preliminary pathology or surgical pathology reports.
Specimen Required
Submit only 1 of the following specimens:

Specimen Type: Tissue

Preferred: Tissue block

Collection Instructions: Submit a formalin-fixed, paraffin-embedded (FFPE) tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used.

Acceptable: Slides

Collection Instructions: Four consecutive, unstained, 5 micron-thick sections placed on positively charged slides, and 1 hematoxylin and eosin-stained slide.

Forms
If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:

Oncology Test Request (T729)

Cardiovascular Test Request (T724)

Specimen Minimum Volume
Two consecutive, unstained, 5 micron-thick sections placed on positively charged slides and 1 hematoxylin and eosin-stained slide

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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

Clinical and Interpretive

Clinical Information
Myxoid/round cell liposarcoma is the second most common subtype of liposarcoma, accounting for more than one-third of all liposarcomas and representing about 10% of all adult soft-tissue sarcomas. Myxoid/round cell liposarcoma is described as a malignant tumor composed of uniform round to oval shaped primitive nonlipogenic mesenchymal cells and a variable number of small signet-ring lipoblasts in a prominent myxoid stroma with a characteristic branching vascular pattern.

A unique chromosome translocation, t(12;16)(q13;p11), resulting in a fusion of the DDIT3 gene (also known as CHOP or GADD153) on chromosome 12 and the FUS gene (also referred to as TLS) on chromosome 16, is the key genetic aberration in myxoid/round cell liposarcoma. More than 90% of myxoid/round cell liposarcoma are
cytogenetically characterized by this translocation. In rare cases, a variant t(12;22)(q13;q12) has been described in which DDIT3 (CHOP) fuses with EWS, a gene highly related to FUS.

Reference Values
An interpretive report will be provided.

Interpretation
A neoplastic clone is detected when the percent of cells with an abnormality exceeds the normal cutoff for the DDIT3 (CHOP) probe.

A positive result is consistent with a subset of myxoid/round cell liposarcoma.

A negative result suggests no rearrangement of the DDIT3 (CHOP) gene region at 12q13. However, this result does not exclude the diagnosis of myxoid/round cell liposarcoma.

Cautions
This test is not approved by the U.S. Food and Drug Administration and it is best used as an adjunct to existing clinical and pathologic information.

Fixatives other than formalin (eg, Prefer, Bouin's) may not be successful for FISH assays; however, nonformalin-fixed samples will not be rejected.

Paraffin-embedded tissues that have been decalcified are generally unsuccessful for FISH analysis. The pathologist reviewing the hematoxylin and eosin-stained slide may find it necessary to cancel testing.

Supportive Data
FISH analysis was performed on 51 formalin-fixed, paraffin-embedded tissue samples including 26 myxoid/round cell liposarcomas and 25 normal soft tissue noncancerous control specimens (from various anatomic locations). The normal controls were used to generate a normal cutoff for this assay. A rearrangement of DDIT3 (CHOP) was identified in 18 of 26 (69%) of myxoid/round cell liposarcoma specimens.

Clinical Reference


Performance

Method Description
The test is performed using a commercially available DDIT3 (CHOP) dual-color, break-apart strategy probe (BAP). Formalin-fixed, paraffin-embedded tissue samples are cut at 5 microns and mounted on positively charged glass slides. The selection of tissue and the identification of target areas on the hematoxylin and eosin (H and E)-stained slide is performed by a pathologist. Using the H and E-stained slide as a reference, target areas are etched with a diamond-tipped etcher on the back of the unstained slide to be assayed. The probe set is hybridized to the appropriate target areas and 2 technologists each analyze 50 interphase nuclei (100 total) with the results expressed as the percent of abnormal nuclei. (Unpublished Mayo method)

**PDF Report**

No

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

Samples processed Monday through Sunday. Results reported Monday through Friday, 8 a.m.-5 p.m. CT.

**Analytic Time**

7 days

**Maximum Laboratory Time**

10 days

**Specimen Retention Time**

Slides and H&E used for analysis are retained by the laboratory in accordance to CAP and NYS requirements. Client provided paraffin blocks and extra unstained slides (if provided) will be returned after testing is complete.

**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 using an analyte specific reagent. Its performance characteristics were 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**

88271x2, 88291 Ą¢â–¬Ą£¢œ DNA probe, each (first probe set), Interpretation and report

88271x2 Ą¢â–¬Ą£¢œ DNA probe, each; each additional probe set (if appropriate)

88271x1 Ą¢â–¬Ą£¢œ DNA probe, each; coverage for sets containing 3 probes (if appropriate)

88271x2 Ą¢â–¬Ą£¢œ DNA probe, each; coverage for sets containing 4 probes (if appropriate)

88271x3 Ą¢â–¬Ą£¢œ DNA probe, each; coverage for sets containing 5 probes (if appropriate)

88274 w/modifier 52 Ą¢â–¬Ą£¢œ Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
**Test Definition: DDITF**

DDIT3, Myxoid Liposarcoma, FISH, Ts

Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)

Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

### LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>DDITF</td>
<td>DDIT3, Myxoid Liposarcoma, FISH, Ts</td>
<td>In Process</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>52155</td>
<td>Result Summary</td>
<td>50397-9</td>
</tr>
<tr>
<td>52157</td>
<td>Interpretation</td>
<td>69965-2</td>
</tr>
<tr>
<td>54585</td>
<td>Result</td>
<td>62356-1</td>
</tr>
<tr>
<td>CG745</td>
<td>Reason for Referral</td>
<td>42349-1</td>
</tr>
<tr>
<td>52158</td>
<td>Specimen</td>
<td>31208-2</td>
</tr>
<tr>
<td>52159</td>
<td>Source</td>
<td>31208-2</td>
</tr>
<tr>
<td>52160</td>
<td>Tissue ID</td>
<td>80398-1</td>
</tr>
<tr>
<td>52161</td>
<td>Method</td>
<td>49549-9</td>
</tr>
<tr>
<td>55027</td>
<td>Additional Information</td>
<td>48767-8</td>
</tr>
<tr>
<td>53830</td>
<td>Disclaimer</td>
<td>62364-5</td>
</tr>
<tr>
<td>52162</td>
<td>Released By</td>
<td>18771-6</td>
</tr>
</tbody>
</table>