

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

Test Id	Reporting Name	Available Separately	Always Performed
_PBCT	Probe, +2	No, (Bill Only)	No
_PADD	Probe, +1	No, (Bill Only)	No
_PB02	Probe, +2	No, (Bill Only)	No
_PB03	Probe, +3	No, (Bill Only)	No
_IL25	Interphases, <25	No, (Bill Only)	No
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No

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

Ordering Guidance

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

- 1. A pathology report is required in order for testing to be performed.** Acceptable pathology reports include working drafts, preliminary pathology or surgical pathology reports.
- 2. A reason for testing must be provided.** If this information is not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.

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

Specimen Type	Temperature	Time	Special Container
Tissue	Ambient (preferred)		
	Refrigerated		

Clinical & 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

1. World Health Organization Classification of Tumours. Pathology and Genetics of Tumours of Soft Tissue and Bone. Edited by CDM Fletcher, K Unni, F Mertens: IARC: Lyon 2002, pp 40-43
2. Meis-Kindblom JM, Sjogren H, Kindblom LG, et al: Cytogenetic and molecular genetic analyses of liposarcoma and its soft tissue simulators: recognition of new variants and differential diagnosis. *Virchows Arch* 2001;439(2):141-51
3. Rabbitts TH, Forster A, Larson R, et al: Fusion of the dominant negative transcription regulator CHOP with a novel gene *FUS* by translocation t(12;16) in malignant liposarcoma. *Nat Genet* 1993 Jun;4(2):175-180
4. Sandberg AA: Updates on the cytogenetics and molecular genetics of bone and soft tissue tumors: liposarcoma. *Cancer Genet Cytogenet* 2004 Nov;155(1):1-24
5. Downs-Kelly E, Goldblum JR, Patel RM, et al: The utility of fluorescence in situ hybridization (FISH) in the diagnosis of

myxoid soft tissue neoplasms. Am J Surg Pathol 2008;32:8-13

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) Performed

Monday through Friday

Report Available

7 to 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 & 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 account representative. 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 US Food and Drug Administration.

CPT Code Information

88271x2, 88291-DNA probe, each (first probe set), Interpretation and report
 88271x2-DNA probe, each; each additional probe set (if appropriate)
 88271x1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
 88271x2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
 88271x3-DNA probe, each; coverage for sets containing 5 probes (if appropriate)
 88274 w/modifier 52-Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
 88274-Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)
 88275-Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
DDITF	DDIT3, Myxoid Liposarcoma, FISH, Ts	101379-6

Result ID	Test Result Name	Result LOINC® Value
52155	Result Summary	50397-9
52157	Interpretation	69965-2
54585	Result	62356-1
CG745	Reason for Referral	42349-1
52158	Specimen	31208-2
52159	Source	31208-2
52160	Tissue ID	80398-1
52161	Method	85069-3
55027	Additional Information	48767-8
52162	Released By	18771-6
53830	Disclaimer	62364-5