MAYO CLINIC LABORATORIES

Test Definition: FUSF

Low-Grade Fibromyxoid Sarcoma (LGFMS), 16p11.2 (FUS or TLS) Rearrangement, FISH, Tissue

Overview

Useful For

Supporting the diagnosis of low-grade fibromyxoid sarcoma when used in conjunction with an anatomic pathology consultation

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 does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered, and the appropriate fluorescence in situ hybridization (FISH) test will be performed at an additional charge.

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.

Appropriate ancillary probes may be performed at consultant discretion to render comprehensive assessment. Any additional probes will have the results included within the final report and will be performed at an additional charge.

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type

Tissue

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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.

Additional Information: Broken or cracked unstained slides will be rejected. Stained slides that were broken and repaired (ie, double mounted) at the client site will not be rejected, as, in most cases, they can still be reviewed under the microscope. However, due to the double mounting, digital scanning cannot be performed.

Forms

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

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

Broken or cracked unstained slides	Reject
Broken or cracked stained slides that cannot be repaired	Reject

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

Low-grade fibromyxoid sarcoma (LGFMS) is a rare malignant soft tissue tumor characterized by a bland fibroblastic spindle cell proliferation arranged in alternating fibrous and myxoid areas, with or without giant collagen rosettes. These tumors are characterized by the chromosome translocation t(7;16)(q33-34;p11), which results in the fusion of *FUS* (also called *TLS*) on chromosome 16 to *CREB3L2* (also called *BBF2H7*) on chromosome 7. Greater than 70% of LGFMS are cytogenetically characterized by this translocation. In rare cases, a variant t(11;16)(p11;p11) has been described in which *FUS* is fused to *CREB3L1* (*OASIS*), a gene structurally related to *CREB3L2*. Testing of *FUS* locus rearrangement should be concomitant with histologic evaluation, and positive results may support the diagnosis of LGFMS.

Reference Values

An interpretative report will be provided.

Interpretation

A neoplastic clone is detected when the percent of cells with an abnormality exceeds the normal cutoff for the *FUS* probe set.

A positive result is consistent with the diagnosis of low-grade fibromyxoid sarcoma (LGFMS).

A negative result suggests that a *FUS* gene rearrangement is not present, but does not exclude the diagnosis of LGFMS.

Cautions

This test is not approved by the US 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) 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.

Broken or cracked unstained slides are not compatible with the instrumentation used in our performing laboratories and will be rejected. Additional material may be requested.

Attempts will be made to repair broken or cracked stained slides; however, if they are unable to be repaired (ie,

shattered in transit), they will be rejected and discarded due to safety concerns.

Supportive Data

FISH analysis was performed on 47 formalin-fixed, paraffin-embedded tissue samples including 13 low-grade fibromyxoid sarcoma (LGFMS) tumors, 8 histologic mimics of LGFMS, and 26 noncancerous control specimens. The normal controls were used to generate a normal cutoff for this assay. Rearrangement of *FUS* was identified in 8 of 13 (62%) LGFMS specimens.

Clinical Reference

1. Fletcher CDM, Unni K, Mertens F: World Health Organization Classification of Tumours. Pathology and Genetics of Tumours of Soft Tissue and Bone. IARC: Lyon 2002, pp 104-105

2. 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

3. Mertens F, Fletcher CD, Antonescu CR, et al: Clinicopathologic and molecular genetic characterization of low-grade fibromyxoid sarcoma, and cloning of a novel *FUS*/*CREB3L1* fusion gene. Lab Invest 2005;85:408-415

4. Vernon SE, Bejarano PA: Low-grade fibromyxoid sarcoma: a brief review. Arch Pathol Lab Med 2006;130:1358-1360

5. Storlazzi CT, Mertens F, et al: Fusion of the FUS and BBF2H7 genes in low grade fibromyxoid sarcoma.Hum Mol Genet. 2003 Sep 15;12(18):2349-58. Epub 2003 Jul 22

Performance

Method Description

This test is performed using a commercially available *FUS* dual-color break-apart strategy probe (BAP). Formalin-fixed paraffin-embedded tissues 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.

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Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
FUSF	FUS (16p11.2), FISH, Ts	101383-8

Result ID	Test Result Name	Result LOINC® Value
52227	Result Summary	50397-9
52229	Interpretation	69965-2
54594	Result	62356-1
CG754	Reason for Referral	42349-1
52230	Specimen	31208-2
52231	Source	31208-2
52232	Tissue ID	80398-1
52233	Method	85069-3
55034	Additional Information	48767-8
52234	Released By	18771-6

53822	Disclaimer	62364-5