MAYO CLINIC LABORATORIES

Test Definition: USPF

USP6 (17p13), Aneurysmal Bone Cyst and Nodular Fasciitis, FISH, Tissue

Overview

Useful For

Supporting the diagnosis of aneurysmal bone cyst or nodular fasciitis

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 consultation. 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.

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 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

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
Container/Tube: Formalin-fixed, paraffin-embedded tumor tissue block
Collection Instructions: Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used

Acceptable: Slides
Specimen Volume: 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 an 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

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

Aneurysmal bone cyst (ABC) is a multicystic and expansile bone tumor of uncertain line of differentiation. *USP6* rearrangements are detectable in approximately 70% of primary ABC and not in other conditions that may simulate ABC histologically, including giant cell tumor of bone, osteosarcoma, osteoblastoma, brown tumor, cherubism, and vascular neoplasms.

Nodular fasciitis (NF) is a self-limited mesenchymal lesion of myofibroblastic differentiation. NFâ€™s rapid growth, rich cellularity, and brisk mitotic activity may lead to a misdiagnosis of sarcoma.

USP6 rearrangements are detectable in 90% of NF but not in other conditions that may simulate NF, including

dermatofibroma, cellular fibrous histiocytoma, fibromatosis, and a large variety of sarcomas.

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 reference range for the *USP6* FISH probe (positive result).

A positive result is consistent with rearrangement of the *USP6* gene locus on 17p13 and supports the diagnosis of aneurysmal bone cyst (ABC) or nodular fasciitis (NF).

A negative result is consistent with no rearrangement of the *USP6* gene locus on 17p13.

However, this result does not exclude the diagnosis of ABC or NF. Rearrangement varies in individual tumors and among different cells in the same tumor.

Cautions

This test is not approved by the FDA 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.

Supportive Data

FISH analysis was performed on 101 samples including 14 primary aneurysmal bone cyst (ABC), 48 nodular fasciitis (NF), and 39 control formalin-fixed paraffin-embedded tissue samples. The normal controls were used to generate a normal cutoff for this assay. A rearrangement of *USP6* was identified in 11 of 14 (77%) and 44 of 48 (92%) NF samples.

Clinical Reference

1. Oliveira AM, Hsi B, Weremowicz S, et al: *USP6* (*Tre2*) fusion oncogenes in aneurysmal bone cyst. Cancer Res 2004 Mar 15;64(6):1920-1923
2. Oliveira AM, Perez-Atayde AR, Inwards CY, et al: *USP6* and *CDH11* oncogenes identify the neoplastic cell in primary aneurysmal bone cysts and are absent in so-called secondary aneurysmal bone cysts. Am J Pathol 2004 Nov;165(5):1773-1780
3. Fletcher CDM, Unni KK, Mertens F: World Health Organization Classification of Tumours. Pathology and Genetics of Tumours of Soft Tissue and Bone. IARC Press, Lyon, France, 2005, pp 48-49
4. Erickson-Johnson MR, Chou MM, Evers BR, et al: Fusion of Non-Muscle Myosin *MYH9* to *USP6* Oncogene in Nodular Fasciitis, USCAP Abstract #39, 2011

Performance

Method Description
The test is performed using a laboratory-developed *USP6* 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-stained slide is performed by a pathologist. Using the hematoxylin and eosin 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 and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It 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
USPF	USP6 (17p13), FISH, Ts	101635-1

Result ID	Test Result Name	Result LOINC® Value
54705	Result Summary	50397-9
54708	Interpretation	69965-2
54707	Result	62356-1
54920	Specimen	31208-2
54710	Source	31208-2
54711	Tissue ID	80398-1
54712	Released By	18771-6
55134	Method	85069-3
55135	Additional Information	48767-8
CG952	Reason for Referral	42349-1
53395	Disclaimer	62364-5