

Overview

Useful For

Confirming the diagnosis of dermatofibrosarcoma protuberans (DFSP)/giant cell fibroblastoma (GCF) and excluding other spindle neoplasms that closely simulate the DFSP histology, including dermatofibroma (benign fibrous histiocytoma), neurofibroma, spindle cell lipoma, and a variety of other benign and malignant spindle cell neoplasms

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 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 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

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

Dermatofibrosarcoma protuberans (DFSP) is a superficial, low-grade sarcoma genetically characterized by the

unbalanced chromosomal translocation t(17;22)(q21;q13), usually in the form of a supernumerary ring chromosome. The product of this chromosomal translocation is the chimeric gene *COL1A1-PDGFB*. Rearrangements of this gene have been detected in approximately 90% of DFSP and its related infantile form, giant cell fibroblastoma, but not in other tumors.

### 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 *PDGFB* FISH probe.

A positive result is consistent with rearrangement/amplification of the *PDGFB* gene locus on 22q13 and supports the diagnosis of dermatofibrosarcoma protuberans (DFSP) or giant cell fibroblastoma (GCF). A negative result is consistent with no rearrangement/amplification of the *PDGFB* gene locus on 22q13. However, this result does not exclude the diagnosis of DFSP or GCF.

The degree of *PDGFB* copy gain/amplification/rearrangement varies in individual tumors and among different cells in the same tumor. It is not currently known if patients with different levels of rearrangement/amplification have the same prognosis and response to therapy.

### 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 49 formalin-fixed paraffin-embedded tissue samples: 24 dermatofibrosarcoma protuberans (DFSP) tumors, 5 giant cell fibroblastoma (GCF) tumors, and 20 non-DFSP/GCF tumors. Normal controls were used to generate a normal cutoff for this assay. Amplification of *PDGFB* was identified in 20 of 24 (83%) and all 5 GCF tumors. Amplification or rearrangement was not identified in the 20 non-DFSP/GCF tumors.

### Clinical Reference

1. Abbott JJ, Erickson-Johnson M, Wang X, et al: Gains of *COL1A1-PDGFB* genomic copies occur in fibrosarcomatous transformation of dermatofibrosarcoma protuberans. *Mod Pathol* 2006 Nov;19(11):1512-1518
2. Labropoulos SV, Fletcher JA, Oliveira AM, et al: Sustained complete remission of metastatic dermatofibrosarcoma protuberans with imatinib mesylate. *Anticancer Drugs* 2005 April;16(4):461-466
3. Macarenco RS, Zamolyi R, Franco MF, et al: Genomic gains of *COL1A1-PDGFB* occur in the evolution of giant cell fibroblastoma into dermatofibrosarcoma protuberans. *Genes Chromosomes Cancer* 2008 Mar;47(3):260-265

## Performance

### Method Description

The test is performed using a laboratory-developed *PDGFB* 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. (Abbott JJ, Erickson-Johnson M, Wang X, et al: Gains of *COL1A1-PDGFB* genomic copies occur in fibrosarcomatous transformation of dermatofibrosarcoma protuberans. *Mod Pathol* 2006 November;19[11]:1512-1518)

### 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

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 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

Test Definition: PDGF

PDGFB (22q13), Dermatofibrosarcoma  
Protuberans/Giant Cell Fibroblastoma, FISH,  
Tissue

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
PDGF	PDGFB (22q13), FISH, Ts	In Process

Result ID	Test Result Name	Result LOINC® Value
54697	Result Summary	50397-9
54700	Interpretation	69965-2
54699	Result	62356-1
54919	Specimen	31208-2
54702	Source	31208-2
54703	Tissue ID	80398-1
54704	Released By	19139-5
CG928	Reason For Referral	42349-1
55130	Method	85069-3
55131	Additional Information	48767-8
53397	Disclaimer	62364-5