Test Definition: PDGF

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

Overview

Useful For

Identifying *PDGFB* gene rearrangements/amplification

Confirming the diagnosis of dermatofibrosarcoma protuberans (DFSP)/giant cell fibroblastoma

Excluding the diagnosis for 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 consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered, and the appropriate fluorescence in situ hybridization (FISH) test will be ordered and performed at an additional charge.

This test includes a charge for the probe application, analysis, and professional interpretation of results for one probe set (2 individual FISH probes). 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

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Specimen

Specimen Type
Tissue

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

Necessary Information
1. A pathology report is required for testing to be performed. If not provided, appropriate testing and/or interpretation may be compromised or delayed. Acceptable pathology reports include working drafts, preliminary pathology, or surgical pathology reports.
2. The following information must be included in the report provided:
-Patient name
-Block number - must be on all blocks, slides, and paperwork
-Date of collection
-Tissue source
3. 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:

Preferred:
Specimen Type: Tissue block
Collection Instructions:
1. Submit a formalin-fixed, paraffin-embedded tumor tissue block. Blocks prepared with alternative fixation methods will be attempted but are less favorable for successful results
2. Provide fixation method used.
Additional Information:
1. Paraffin-embedded specimens can be from any anatomic location (skin, soft tissue, lymph node, etc).
2. Bone specimens that have been decalcified will be attempted for testing, but the success rate is approximately 50%.

Acceptable:
Specimen Type: Tissue slides
Slides: 1 Hematoxylin and eosin-stained and 2 unstained
Collection Instructions: Submit 2 consecutive unstained, positively charged, unbaked slides with 5 micron-thick sections of the tumor tissue and 1 slide stained with hematoxylin and eosin.
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 an Oncology Test Request (T729) with the specimen.

Specimen Minimum Volume

Slides: 1 Hematoxylin and eosin stained and 2 unstained

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

Dermatofibrosarcoma protuberans is a superficial, low-grade sarcoma genetically characterized by the unbalanced chromosomal translocation t(17;22)(q21.33; q13.1), usually in the form of a supernumerary ring chromosome. The product of this chromosomal translocation is the *COL1A1::PDGFB* chimeric gene. 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

PDGFB will be clinically interpreted as positive, negative, or equivocal.

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

A positive result is consistent with rearrangement/amplification of the *PDGFB* gene. A positive result may support a

diagnosis of dermatofibrosarcoma protuberans (DFSP) or giant cell fibroblastoma (GCF). The significance of this finding is dependent on the clinical and pathologic features.

A negative result suggests a *PDGFB* gene rearrangement/amplification is not present. A negative 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 U.S. Food and Drug Administration and is best used as an adjunct to existing clinical and pathologic information.

This fluorescence in situ hybridization (FISH) assay does not rule out other chromosome abnormalities.

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

Paraffin-embedded tissues that have been decalcified may not be successful for FISH analysis. The success rate of FISH studies on decalcified tissue is approximately 50%, but FISH will be attempted if sufficient tumor is present for analysis.

Fluorescence in situ hybridization studies will be attempted if sufficient tumor is present for analysis. The pathologist reviewing the hematoxylin and eosin-stained slide may find it necessary to cancel testing if insufficient tissue/tumor is available for testing.

If no FISH signals or a lack of sufficient tumor tissue are observed post-hybridization, the case will be released indicating a lack of FISH results.

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.

Clinical Reference

1. Abbott JJ, Erickson-Johnson M, Wang X, Nascimento AG, Oliveira AM. Gains of *COL1A1-PDGFB* genomic copies occur in fibrosarcomatous transformation of dermatofibrosarcoma protuberans. Mod Pathol 2006;19(11):1512-1518
2. Labropoulos SV, Fletcher JA, Oliveira AM, Papadopoulos S, Razis ED. Sustained complete remission of metastatic dermatofibrosarcoma protuberans with imatinib mesylate. Anticancer Drugs 2005;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;47(3):260-265
4. WHO Classification of Tumours Editorial Board, eds. Soft Tissue and Bone Tumours, 5th ed. Lyon: IARC Press; 2020.

WHO Classification of Tumours, Volume 3.

Performance

Method Description

The test is performed using a laboratory-developed PDGFB dual-color, break-apart strategy fluorescence in situ hybridization probe set (BAP). 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 are performed by a pathologist. Using the H and E-stained slide as a reference, target areas are etched with a diamond-tipped engraving tool on the back of the unstained slide to be assayed. Each probe set is hybridized to the appropriate target areas, as indicated on the H and E, and 100 interphase nuclei are scored within the targeted areas. The results are 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 used for analysis are retained by the laboratory in accordance with regulatory 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.

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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
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