

TGFBR3 (1p22), OGA (10q24) Rearrangement, FISH, Tissue

Overview

Useful For

Supporting the diagnosis of pleomorphic hyalinizing angiectatic tumor, or hemosiderotic fibrolipomatous tumors associated with rearrangement of the *OGA* and/or *TGFBR3* gene when used in conjunction with an anatomic pathology consultation

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_PBCT	Probe, +2	No	No
_PADD	Probe, +1	No	No
_PB02	Probe, +2	No	No
_PB03	Probe, +3	No	No
_IL25	Interphases, <25	No	No
_1099	Interphases, 25-99	No	No
_1300	Interphases, >=100	No	No

Testing Algorithm

This test includes a charge for the probe application, analysis, and professional interpretation of results for 2 probe sets (4 individual fluorescence in situ hybridization [FISH] probes). No analysis charges will be incurred if an insufficient number of representative cells are available for analysis.

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type

Tissue

Ordering Guidance

This test does not include a pathology consultation. If a pathology consultation is requested, order PATHC / Pathology Consultation, and appropriate testing will be added at the discretion of the pathologist and performed at an additional charge.

Shipping Instructions



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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: Submit a formalin-fixed, paraffin-embedded tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; 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 6 unstained

Collection Instructions: Submit 1 slide stained with hematoxylin and eosin and 6 consecutive unstained, positively charged, unbaked slides with 5 micron-thick sections of the tumor tissue.

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

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		



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Clinical & Interpretive

Clinical Information

Chromosomal rearrangement involving the *OGA* (previously *MGEA5*) gene or the *TGFBR3* gene have recently been associated with both pleomorphic hyalinizing angiectatic tumor of soft parts and hemosiderotic fibrolipomatous tumor. Either one or both rearrangements may be present and support a diagnosis in the proper clinical and pathologic context. Rearrangement of one or both genes may be present in other neoplastic processes and is not diagnostic in isolation. These rearrangements are unusual in that they do not result specific RNA transcript products and therefore are often not detectable through many next-generation sequencing approaches but can typically be detected by fluorescence in situ hybridization testing.

Reference Values

An interpretive report will be provided.

Interpretation

The result is considered positive when the percent of cells with separation of the *OGA* and/or *TGFBR3* FISH probes with an abnormality exceeds the normal cutoff for the OGA and/or TGFBR3 FISH probe set.

A positive result suggests rearrangement of the OGA and/or TGFBR3 gene and likely reflects OGA and/or TGFBR3 fusion with a partner gene. The significance of this finding is dependent on the clinical and pathologic features.

The result is considered negative when the percent of cells with separation of the *OGA* and/or *TGFBR3* FISH probes does not exceed the normal cutoff for the *OGA* or *TGFBR3* FISH probe set.

A negative result does not exclude the presence of a OGA or TGFBR3 rearrangement.

Rearrangement of the OGA or TGFBR3 gene may be present in other neoplastic processes.

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's) may not be successful for fluorescence in situ hybridization (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%.

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

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



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Clinical Reference

- 1. WHO Classification of Tumours Editorial Board. Soft Tissue and Bone Tumours. 5th ed. IARC; 2020. WHO Classification of Tumours Series. Vol. 3, 280-283
- 2. Zreik RT, Carter JM, Sukov WR, et al. TGFBR3 and MGEA5 rearrangements are much more common in 'hybrid' hemosiderotic fibrolipomatous tumor-myxoinflammatory fibroblastic sarcomas than in classical myxoinflammatory fibroblastic sarcomas: a morphological and fluorescence in situ hybridization study. Hum Pathol. 2016;53:14-24

Performance

Method Description

The test is performed using laboratory-developed *TGFBR3* (1p22) and *OGA* (10q24) dual-color break-apart strategy probes. 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 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. The probe set is hybridized to the appropriate target areas, and 2 technologists independently 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 with regulatory requirements. Client provided paraffin blocks and extra unstained slides 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 <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.



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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)

88271-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
TGOGF	TGFBR3, OGA, FISH, Ts	106436-9

Result ID	Test Result Name	Result LOINC® Value
621842	Result Summary	50397-9
621843	Interpretation	69965-2
621844	Result	62356-1
GC145	Reason for Referral	42349-1
621845	Specimen	31208-2
621846	Source	31208-2
621847	Tissue ID	80398-1
621848	Method	85069-3
621849	Additional Information	48767-8
621850	Disclaimer	62364-5
621851	Released By	19139-5