MAYO CLINIC LABORATORIES

Test Definition: TFEBF

Renal Cell Carcinoma, 6p21.1 (TFEB) Rearrangement, FISH, Tissue

Overview

Useful For

Identifying *TFEB* gene rearrangements

Supporting the diagnosis of renal cell carcinoma when used in conjunction with an anatomic pathology consultation

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No
_IL25	Interphases, <25	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
_PBCT	Probe, +2	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

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 (FFPE) tumor tissue block. Blocks prepared with alternative fixation methods will be attempted but are less favorable for successful results by FISH testing.
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 4 unstained

Collection Instructions: Submit 4 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

The gene *TFEB* is often altered in renal cell carcinoma (RCC). Fluorescence in situ hybridization analysis allows for the detection of rearrangement of the *TFEB* gene region and can aid in the diagnosis of RCC.

Reference Values

An interpretive report will be provided.

Interpretation

TFEB 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 TFEB probe set.

A positive result is consistent with rearrangement of the *TFEB* gene and likely reflects *TFEB* fusion with a partner gene. A positive result of TFEB suggests promotor substitution caused by structural alterations of the *TFEB* gene region at 6p21. The significance of this finding is dependent on the clinical and pathologic features.

A negative result suggests a *TFEB* gene rearrangement is not present. A negative result does not exclude the diagnosis of renal cell carcinoma.

Cautions

This test is not approved by the US 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.

FISH 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. Argani P, Yonescu R, Morsberger L, et al. Molecular confirmation of t(6;11)(p21;q12) renal cell carcinoma in archival paraffin-embedded material using a break-apart TFEB FISH assay expands its clinicopathologic spectrum. Am J Surg Pathol. 2012;36(10):1516-1526
2. Argani P, Cheville J, Ladanyi M. MiT family translocation renal cell carcinomas. In: Moch H, Humphrey PA, Ulbright TM, Reuter VE. WHO Classifications of Tumours of Urinary System and Male Genital Organs. 4th ed. IARC Press, 2016, 33-34

Performance

Method Description

This test uses a laboratory developed TFEB dual-color, break-apart strategy fluorescence in situ hybridization probe set. 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 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
88271 x 2, 88291-DNA probe, each (first probe set), Interpretation and report
88271 x 2-DNA probe, each; each additional probe set (if appropriate)
88271 x 1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
88271 x 2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
88271 x 3-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
TFEBF	TFEB, 6p21.1, FISH	95780-3

Result ID	Test Result Name	Result LOINC® Value
92350	Result Summary	50397-9
92351	Interpretation	69965-2
92352	Result	62356-1
GC002	Reason for Referral	42349-1
92353	Specimen	31208-2
92354	Source	31208-2

92355	Tissue ID	80398-1
92356	Method	85069-3
92357	Additional Information	48767-8
92358	Disclaimer	62364-5
92359	Released By	18771-6