

## Overview

### Useful For

Identifying *TFEB* gene rearrangements in patients with renal cell carcinoma (RCC)

### 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 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

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

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  
Container/Tube: Formalin-fixed, paraffin-embedded tumor tissue block

Specimen Type: Slides  
Specimen Volume: 4 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

The *TFEB* gene may be altered in some patients with renal cell carcinoma (RCC). Identification of rearrangement of the *TFEB* gene region by FISH analysis can aid in the diagnosis of RCC.

Reference Values

An interpretive report will be provided.

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**Interpretation**

A positive result with the *TFEB* probe is detected when the percent of cells with an abnormality exceeds the normal cutoff for the probe set. A positive result of TFEB suggests promotor substitution caused by structural alterations of the *TFEB* gene region at 6p21.1. A negative result suggests no structural alterations of the locus.

**Cautions**

This test is not approved by the FDA and should be used as an adjunct to existing clinical and pathologic information.

Fixatives other than formalin (eg, Prefer, Bouin) may not be successful for FISH assays. Although FISH testing will not be rejected due to nonformalin fixation, results may be compromised.

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 27 formalin-fixed paraffin-embedded specimens including 25 renal cell carcinoma (RCC) tissue samples, 2 RCC specimens from Johns Hopkins known positive for *TFEB* rearrangement, and 25 noncancerous control specimens. The normal controls were used to generate the normal cutoff values. Structural alterations resulting in the rearrangement of the *TFEB* gene region were identified and results correlated with pathology findings.

**Clinical Reference**

- 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 Oct;36(10):1516-1526
- Argani P, Cheville J, Ladanyi M: MiT family translocation renal cell carcinomas. *In* WHO Classifications of Tumours of Urinary System and Male Genital Organs. Fourth edition. Edited by H Moch, PA Humphrey, TM Ulbright, VE Reuter VE. Lyon, France, IARC Press, 2016, 33-34

**Performance****Method Description**

This test uses a laboratory developed *TFEB* (6p21.1) dual-color break-apart probe (BAP) strategy. 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 etcher on the back of the unstained slide to be assayed. The probes are hybridized to the appropriate target areas and 2 technologists analyze each probe set. Using the *TFEB* probe set, each technologist analyzes 50 interphase nuclei (100 total) and 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 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

- 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

Test Definition: TFEBF

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

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