

TEST ID: TFEBF

RENAL CELL CARCINOMA, 6P21.1 (*TFEB*) REARRANGEMENT, FISH, TISSUE

USEFUL FOR

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

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 FISH test will be ordered and 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.

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.

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.

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.

REFERENCE VALUES

An interpretative report will be provided.

ANALYTIC TIME

5 days

SPECIMEN REQUIRED

Submit only 1 of the following specimens:

Type

Tissue

Container/Tube

Formalin-fixed, paraffin-embedded tumor tissue block

Type

Slides

Container/Tube

4 Consecutive, unstained, 5 micron-thick sections placed on positively charged slides and 1 hematoxylin and eosin-stained slide

RENAL CELL CARCINOMA, 6P21.1 (TFEB) REARRANGEMENT, FISH, TISSUE**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 TFEF FISH assay expands its clinicopathologic spectrum. Am J Surg Pathol 2012 Oct;36(10):1516-1526
2. 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