



Empire Genomics Probe Used in Characterizing a Case of TFEB Renal Cell Carcinoma

Background

t(6;11)(p21;q12)/TFEB gene fusion-associated renal cell carcinoma, or TFEB RCC, is a recently recognized renal cell carcinoma caused by the formation of Alpha-TFEB fusion genes. Identification of TFEB rearrangements via FISH analysis is a crucial step in diagnosis of the disease.

Objectives

In this study, appropriate tools and techniques were utilized to report a rare case of t(6;11)(p21;q12)/TFEB gene fusion-associated renal cell carcinoma and its clinicopathologic features.

Approach

Radical left nephrectomy was performed on the patient and gross findings were reported. Microscopic findings regarding the excised tumor were also reported, including its composition of medium and large epithelioid cells in a solid nesting or acinous cellular arrangement. Immunohistochemical staining of the tumor was used to identify genes for which the tumor cells tested positive. A TFEB gene break-apart probe manufactured by Empire Genomics was used for FISH testing. The TFEB probe was marked green on the centromeric side and orange on the telomeric side, and when the distance between the orange and green signals was greater than the size of one fusion signal, signal break-apart was deemed to have occurred.

Results

The TFEB gene break-apart probe from Empire Genomics was used in FISH analysis to help characterize a case of TFEB renal cell carcinoma. In this case, a break-apart signal indicating rearrangement of the TFEB gene was determined by FISH, a crucial diagnostic tool in the characterization of this disease.

t(6;11)(p21;q12)/TFEB gene fusion-associated renal cell carcinoma: a case report and review of literature

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Lead Organization

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Diseases

- Renal Cell Carcinoma

Biomarkers Mentioned

- E-cadherin
- HMB45
- Melan-A
- AE1/AE3
- Vimentin
- RCC
- CK19
- P504S
- CD10
- CD117
- CD34
- S-100
- EMA
- Desmin
- Cam5.2
- TFE3
- CK7
- Ki-67