Kidney: Renal cell carcinoma with t(X;1)(p11;q21) PRCC/TFE3

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Abstract

Review on Renal cell carcinoma with t(X;1)(p11;q21) PRCC/TFE3, with data on clinics, and the genes involve

Keywords
Renal cell carcinoma; chromosome X; chromosome 1; PRCC; TFE3; MiT family

Classification

Xp11 translocation renal cell carcinoma (RCCs) harbor gene fusions involving TFE3 transcription factor. The t(6;11) RCCs harbor a specific MALAT1 (Alpha) - TFEB gene fusion. TFEB and TFE3 belong to the same MiT subfamily of transcription factors.

Because of similarities at the clinical, morphologic, immunohistochemical, and genetic levels, the Xp11 translocation RCCs and t(6;11) RCCs are currently grouped together under the category of MiT family translocation renal cell carcinoma.

Clinics and pathology

Disease
Renal cell carcinoma.

Phenotype / cell stem origin

Xp11 translocation renal cell carcinoma. t(X;1) (p11;q21) PRCC/TFE3 is found in Xp11 translocation renal cell carcinoma.
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**Epidemiology**
Approximately 40 reported cases, median age 20 yrs (range 2-69); sex ratio: 17M/20F. A subset of cases has been associated with prior exposure to cytotoxic chemotherapy.

**Pathology**
PRCC frequently have papillary architecture with clear cytoplasm and abundant psammoma bodies. Other cases are nested and mimic clear cell RCC.

**Treatment**
Radical nephrectomy.

**Prognosis**
Similar to that of other papillary renal cell carcinoma.

**Cytogenetics**

**Probes**
TFE3 and PRCC/TFE3

**Additional anomalies**
+17.

**Genes involved and proteins**

**TFE3 (transcription factor E3)**
- **Location**: Xp11.23
- **Protein**: Transcription factor; binds to the immunoglobulin enhancer.

**PRCC (papillary renal cell carcinoma)**
- **Location**: 1q23.1
- **Protein**: 491aa; widely expressed; proline rich.

**Result of the chromosomal anomaly**

**Hybrid Gene**
- **Description**: 5’ TFE3 - 3’ PRCC and 5’ PRCC - 3’ TFE3
- **Detection**: positional cloning; screening in a tumor cDNA library and hybridization with TFE3

**Fusion Protein**
- **Description**: PRCC/TFE3 fusion protein includes the transcription activating, helix-loop-helix and leucine-zipper domains of TFE3; the TFE3/PRCC fusion protein (513 amino acids) is also expressed.

**References**


This article should be referenced as such: