

Solid Tumour Section

Short Communication

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

t(X;1) (p11;q21) PRCC/TFE3 is found in Xp11 translocation renal cell carcinoma.

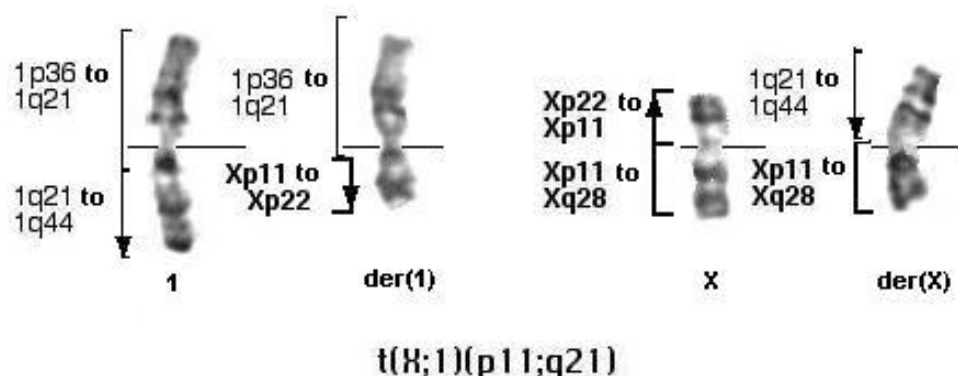


Figure 1 t(X;1)(p11;q21) (G banding) - Courtesy Francois Desangles.

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 others 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.

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