Solid Tumour Section
Mini Review

t(X;1)(p11.2;q21.2) in renal cell carcinoma
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Identity

Classification
Renal cell carcinoma (RCC) are classified into two main clinico-pathologic entities (chromophobic/oncocytic entity excluded): Clear-cell RCC (also called non-papillary RCC, found in 80% of cases); Chromophilic RCC (also called papillary RCC, in 15-20% of cases).

Clinics and pathology

Disease
Renal cell carcinoma.

Phenotype stem cell origin
t(X;1) is found in papillary renal cell carcinoma (WHO classification).

Epidemiology
12 reported cases, herein reviewed; mean age 16 yrs (range 1-30); sex ratio: 7M/5F (female cases are found, contrarily to what has previously been suspected).

Pathology
t(X;1) appears to delineate a distinct subgroup of chromophilic papillary renal cell carcinoma with clear cell features.

Treatment
Radical nephrectomy.

Prognosis
Similar to that of others papillary renal cell carcinoma i. e. overall 5-yr survival rate around 85%.

Cytogenetics

Probes
TFE3 and PRCC/TFE3.

Cytogenetics Molecular
+7, +17.

Variants
t(X;1)(p11.2;q21) (G-banding) - Courtesy François Desangles.

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Genes involved and Proteins

**TFE3**
*Location:* Xp11.2
*Protein*
Transcription factor; binds to the immunoglobulin enhancer.

**PRCC**
*Location:* 1q21.2
*Protein*
491 aa; widely expressed; proline rich.

Result of the chromosomal anomaly

**Hybride Gene**
*Description*
5' TFE3 - 3' PRCC and 5' PRCC - 3' TFE3.

**Detection protocol**
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.

To be noted
We have had a t(X;1) case in a non-papillary renal cell carcinoma, suggesting that t(X;1) is not restricted to the papillary subtype of RCC (unpublished).

References


This article should be referenced as such: