Kidney: t(X;1)(p11.2;p34) in renal cell carcinoma

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Identity

Note: Must not be confused with the t(X;1)(p11.2;q21), also found in renal cell carcinoma.

Classification

Renal cell carcinoma (RCC) are classified into two main clinico-pathologic entities
- 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

t(X;1)(p11.2;p34) is found in very rare (n<5) cases of papillary renal cell carcinoma.

Phenotype / cell stem origin

May not be restricted to the papillary subtype of renal cell carcinoma.

Genes involved and proteins

TFE3
Location
Xp11.2
Protein Contains a transcriptional activation domain, a helix-loop-helix, and a leucine zipper; member of the basic helix-loop-helix family (b-HLH) of transcription factors.

PSF
Location
1p34
Protein Contains RNA binding domains; involved in pre-m RNA splicing; form complexes with DNA topoisomerase I.

Result of the chromosomal anomaly

Hybrid Gene

Description
5' PSF - 3' TFE3.

Fusion Protein

Description
N-term PSF and most of it fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); no TFE3-PSF reciprocal transcript, as the der(X) t(X;1) is missing; the normal TFE3 transcript is found.

References


This article should be referenced as such: