Solid Tumour Section
Short Communication

Kidney: Primary renal ASPSCR1-TFE3 t(X;17)(p11;q25) tumor
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Classification

Low power image of t(X;17) renal carcinoma section showing characteristic alveolar pseudopapillary architecture with dark staining psammomatous calcifications - Courtesy Matt Burtelow and Charles D. Bangs.
Clinics and pathology

Disease
Previously diagnosed as papillary renal cell carcinoma, they share cytogenetic features with alveolar soft part sarcoma, and they exhibit pathological characteristics of one or the other tumours, or intermediate features.

Epidemiology
Mainly found in young patients (1.5 yr to 17 yrs in a series).

Treatment
Surgery.

Prognosis
Relatively indolent clinical course.
Cytogenetics

Balanced t(X;17)(p11.2;q25). This is in contrast with what is found in the alveolar soft part sarcoma where the translocation t(X;17)(p11.2;q25) involves the same breakpoints and the same genes, but is found unbalanced in most, if not all, the cases.

Genes involved and proteins

TFE3
Location
Xp11
DNA / RNA
8 exons
Protein
Transcription factor; member of the basic helix-loop-helix family (b-HLH) of transcription factors primarily found to bind to the immunoglobulin enhancer muE3 motif.

ASPCRN1
Location
17q25
Protein
476 amino acids; contains an UBX domain.

Result of the chromosomal anomaly

Hybrid Gene
Description
5' ASPSCR1-3' TFE3: ASPSCR1 is fused in frame to TFE3 exon 3 or 4 the reciprocal 5' TFE3 - 3' ASPSCR1 may or may not be transcribed.

Fusion Protein
Description
NH2 term from ASPSCR1, fused to the C term of TFE3, including the activation domain, the helix-loop-helix, and the leucine zipper from TFE3.

References

This article should be referenced as such: