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Solid Tumour Section

Short Communication

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

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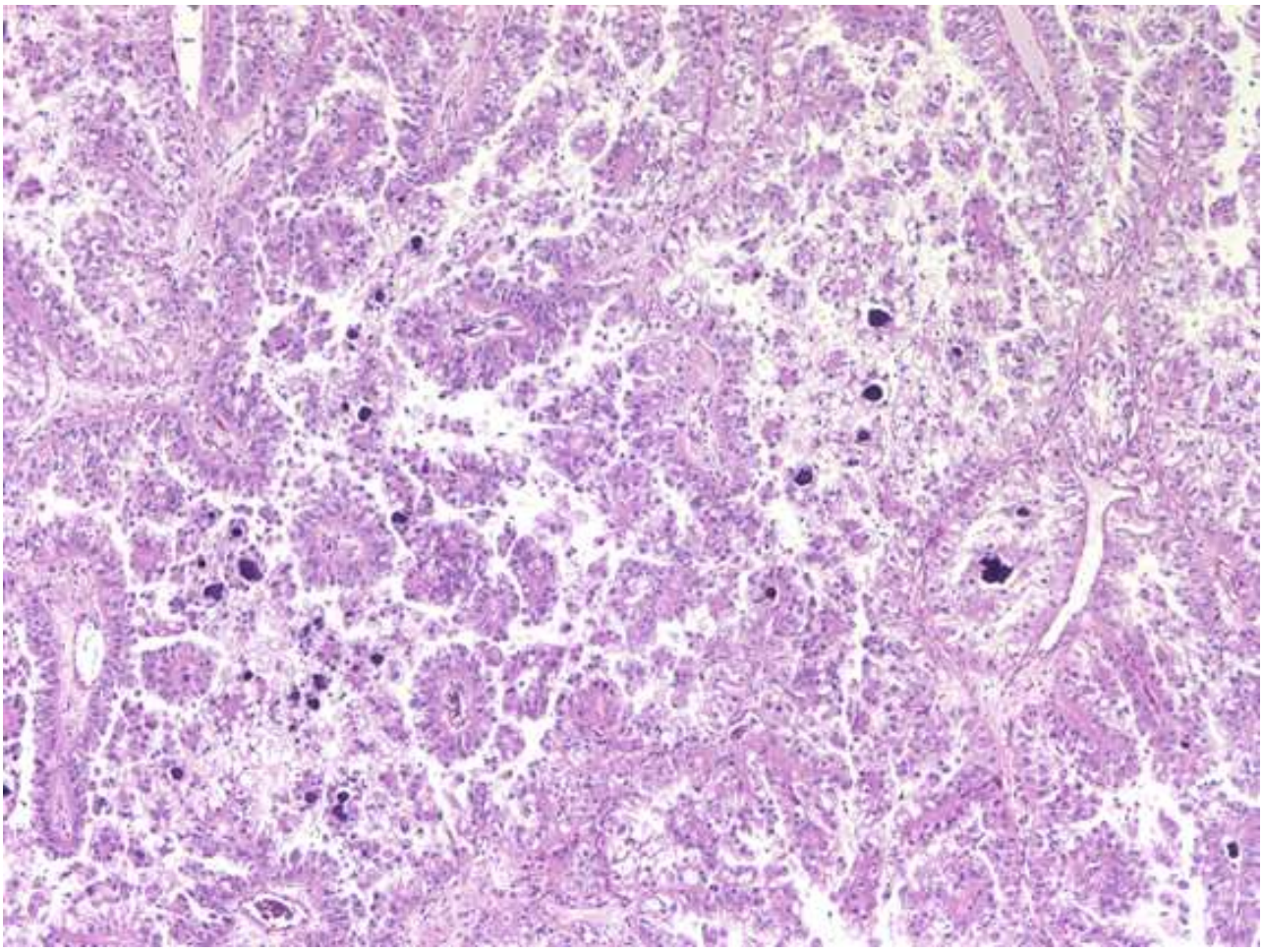
Classification



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 exibit pathological characteristics of one or the other tumours, or intermediate features.

Epidemiology

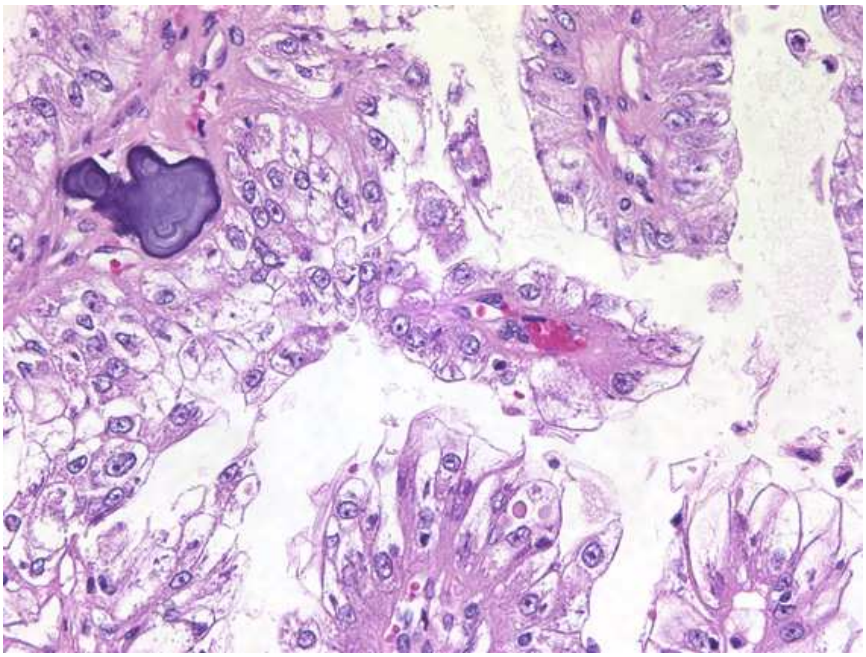
Mainly found in young patients (1.5 yr to 17 yrs in a series).

Treatment

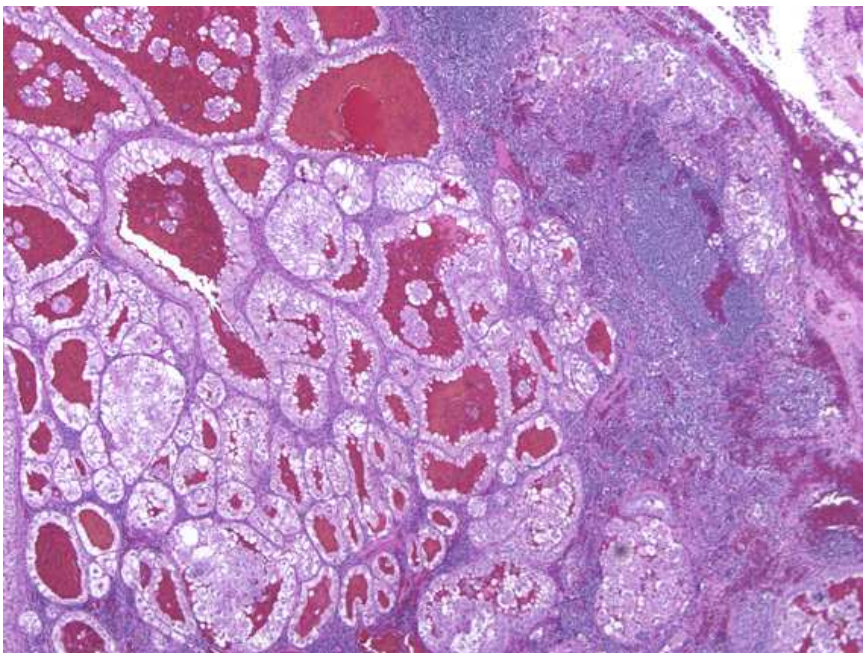
Surgery.

Prognosis

Relatively indolent clinical course.

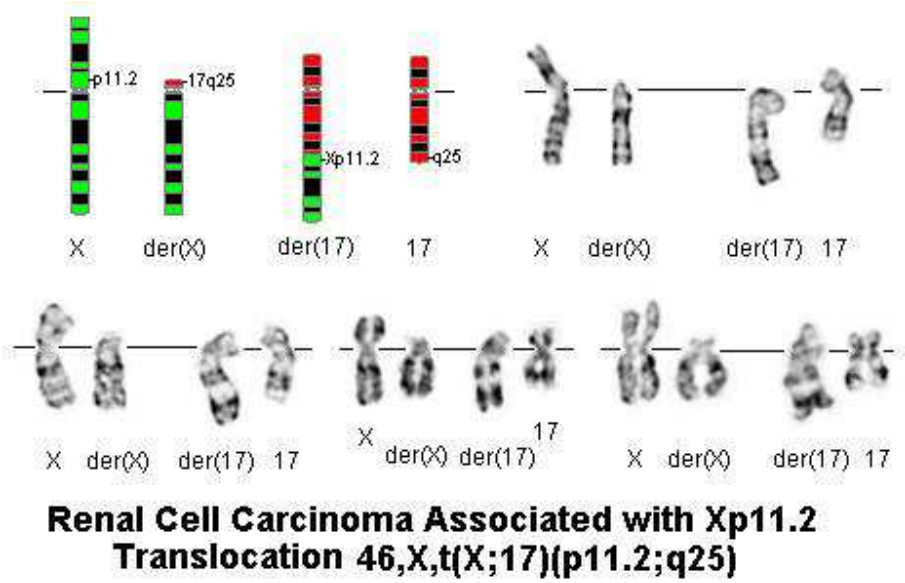


High-power image showing typical clear cells with voluminous cytoplasm and a large psammoma body - Courtesy Matt Burtelow and Charles D. Bangs.



Hilar lymph node section showing replacement of normal nodal structure by nests of metastatic renal cell carcinoma - Courtesy Matt Burtelow and Charles D. Bangs.

Cytogenetics



t(X;17)(p11;q25) G- banding: - Courtesy Charles D. Bangs.

Cytogenetics Morphological

Balanced t(X;17)(p11.2;q25). This is in contrast with what is found in the alveolar soft part sarcoma where the tranlocation 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 enchancer muE3 motif.

ASPSCR1

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

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Argani P, Antonescu CR, Illei PB, Lui MY, Timmons CF, Newbury R, Reuter VE, Garvin AJ, Perez-Atayde AR, Fletcher JA, Beckwith JB, Bridge JA, Ladanyi M. Primary renal neoplasms with the ASPL-TFE3 gene fusion of alveolar soft part sarcoma: a distinctive tumor entity previously included among renal cell carcinomas of children and adolescents. Am J Pathol. 2001 Jul;159(1):179-92

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