

Solid Tumour Section

Short Communication

Soft tissue tumors: Alveolar soft part sarcoma

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Clinics and pathology

Embryonic origin

The histogenesis of this tumour is still unknown, despite immunohistochemistry and electron microscopy studies.

Epidemiology

Often occurs in the young adult, more frequently in females.

Clinics

Involve the muscles and soft tissues of the lower extremities in adults (in more than half cases), the head and neck regions in the child, but it can also have extra muscular localizations, such as the female genital tract, the trunk, or the retroperitoneum; brain metastases are frequent.

Pathology

Secretory process with the formation of cytoplasmic crystals can be seen with electron microscopy.

Treatment

Aggressive surgical excision.

Prognosis

Relatively indolent clinical course. Overall survival of adult patients without metastases reaches 87% at 5 years, and that of adult patients with metastases is 20%, with a median survival of 40 months. Pediatric cases have a better prognosis, with a 5 years survival of 80%

for all cases included, reaching 91% in cases without metastases.

Cytogenetics

Cytogenetics Morphological

der(X)t(X;17)(p11;q25) is consistently involved; it implicates: 1- the formation of a hybrid gene at the breakpoint, and also, 2- gain in Xp11-pter sequences, and loss of heterozygosity in 11q25-qter, with possible implications.

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.

ASPSCR1

Location

17q25

Protein

476 amino acids; contains an UBX domain.

Result of the chromosomal anomaly

Hybrid Gene

Description

5' ASPSCR1-3' TFE3; the reciprocal 5' TFE3 - 3' ASPSCR1 is most often absent. ASPSCR1 is fused in frame to TFE3 exon 3 or 4.

Fusion Protein

Description

234 NH2 term amino acids from ASPSCR1, fused to the 280 or 315 C term amino acids from TFE3, including the activation domain, the helix-loop-helix, and the leucine zipper from TFE3.

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