Solid Tumour Section
Mini Review

Soft tissue tumors: Extraskeletal myxoid chondrosarcoma

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Clinics and pathology

Disease
Malignant tumour of soft tissue origin, distinct from the primary skeletal chondrosarcoma with myxoid alteration.

Epidemiology
It is a rare tumour: 2.3% of soft tissue sarcomas in a Japanese series; mean ages reported in various series range from 46 to 57 years, this tumour being exceptional in children and adolescents; males are affected about twice as often as females.

Clinics
Location: deep soft tissues of the lower extremities in about 75% of the cases, especially the thigh, the popliteal fossa, and the buttock; occasionally, a bone involvement may exist, as a minor component.

Pathology
Macroscopic findings: the tumour presents as lobulated or multinodular mass, generally well circumscribed by a distinct fibrous capsule. The size of the tumour at the time of diagnosis may vary from 1 to about 20 cm (mean size: about 7 cm).
Histology: typically, tumour nodules are composed of round or slightly elongated cells, with features of chondroblasts, separated by mucoid substance; differentiated cartilage cells are rare; histological diagnosis may be very difficult, especially in highly cellular forms devoid of myxoid matrix.
Tumour cells generally show positivity for vimentin, S-100 protein, occasionally for EMA, and negativity for cytokeratin.

Treatment
Treatment: surgical excision, with adjuvant chemotherapy in case of lymph nodes or metastasis.

Cytogenetics

Cytogenetics Morphological
Cytogenetic studies have demonstrated the presence of a recurrent translocation t(9;22)(q22;q12); it results in the fusion of the EWSR1 gene on chromosome 22 with TEC (or CHN) gene on chromosome 9.
Recently, a variant translocation t(9;17)(q22;q11) has been identified, fusing the gene TEC to gene TAF2N (TAFII68, or RBP56).

Genes involved and proteins

TEC
Location
9q22
DNA / RNA
Transcripts: 2.6 kb and 3.7 kb.
Protein
Signaling mediator; activate the c-fos promoter; role in growth and differentiation processes of hematopoietic tissues.

EWSR1
Location
22q12
DNA / RNA
17 exons; 2.4 kb mRNA.
Protein
RNA-binding protein; transcription repressor.

**TAF2N**

Location
17q11.1-q11.2

DNA / RNA
16 exons; alternative splicing; 2.2 bp mRNA.

Protein
RNA-binding protein; part of the TFIID and RNA polymerase II complex.

### Result of the chromosomal anomaly

**Fusion Protein**

**Description**
The EWS/TEC(CHN) gene fusion encodes a fusion protein in which the C-terminal RNA-binding domain of EWS is replaced by the entire TEC protein. TEC is a member of the steroid/thyroid receptor gene superfamily; the EWS/TEC fusion protein is a potent transcriptional activator.
The TAF2N/TEC fusion, in which exon 6 of TAF2N(TAFII68, or RBP56) is fused to the entire coding region of TEC, is structurally and functionally very similar to the EWS/TEC fusion.

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