Soft tissue tumors: t(X;20)(p11.23;q13.33) in biphasic synovial sarcoma

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Clinics and pathology

Disease
Biphasic Synovial Sarcoma

Pathology
The excised tumor consisted of a single 3-cm nodulus with relatively well-defined borders and a grey cut surface. No necrosis was seen. In histological sections stained with H and E, the tumor was mainly composed of uniform, closely packed spindle cells, with a high nuclear/cytoplasmic ratio and finely dispersed chromatin. The tumor cells were arranged in fascicles and whorls, with small foci of calcification. The mitotic activity was low. In addition, small and irregular glandular formations lined by flattened cuboidal cells were noted in the peripheral parts of the tumor, supporting the diagnosis of biphasic synovial sarcoma. These cells, as well as many spindle cells, expressed cytokeratins and epithelial membrane antigen (EMA).

Treatment
The patient was given postoperative chemo- and radiotherapy.

Evolution
Four months postoperatively, the patient is alive without any sign of local recurrence or metastatic disease.

Cytogenetics

Cytogenetics morphological
48,XY,+mar1,+mar2[5]/47,idem, dic(19;22)(q13;q13)[3].

Cytogenetics molecular
With a pool of SSX BAC probes, one and two signals were seen on mar1 and mar2, respectively. On the latter marker chromosome, one of the signals co-localized with the signal from RP5-1005F21, suggesting the presence of a fusion gene between SS18L1 and one of the SSX genes.

Probes
RP5-1005F21 (SS18L1), RP11-552E4 and RP11-344N17 (pool of SSX genes).
**Genes involved and Proteins**

**SS18L1**

Location: 20q13.33  
Note: Paralogous gene to SS18 (18q11.2).

DNA/RNA  

**SSX1**

Location: Xp11.23  
Note: Gene belonging to the SSX gene family.

DNA/RNA  

Protein  
188 amino acids (Q16384).

**Result of the chromosomal anomaly**

**Hybride Gene**

Description  
Nucleotide 1216 (exon 10) of SS18L1 (Accession Number XM_037202) was fused in-frame with nt 422 (exon 6) of SSX1 (Accession Number X86174).

**Fusion protein**

Description  
In the putative SS18L1/SSX1 chimeric protein, the last 8 amino acid residues of the SS18L1 protein are replaced by 78 amino acids from the COOH-terminal part of SSX1. By analogy with what is presumed to be the case for the SS18/SSX fusion protein, SS18L1/SSX1 is likely to show an altered transcriptional pattern, with the COOH-terminal SSX domain redirecting the SS18L1 activation domain to new target promoters.

**References**


*This article should be referenced as such: Storlazzi CT, Mertens F. Soft tissue tumors: t(X;20)(p11.23;q13.33) in biphasic synovial sarcoma. Atlas Genet Cytogenet Oncol Haematol. 2007;11(4):355-356.*