Atlas of Genetics and Cytogenetics
in Oncology and Haematology

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# Solid Tumour Section

## Mini Review

# 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

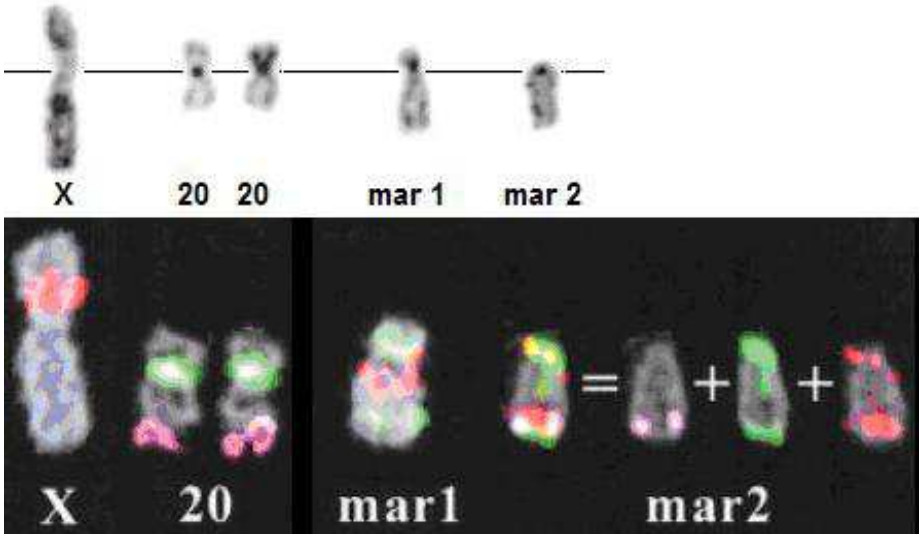
48,XY,+mar1,+mar2[5]/47,idem, dic(19;22)(q13;q13)[3].

### 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).




Top: Partial karyotype showing G-banded chromosomes X, 18, 20 and markers 1 and 2.
Bottom: FISH cohybridization using a pool of RP11-552E4 and RP11-344N17 (red), RP5-1005F21 (purple), and pZ20 (green) as probes for chromosomes X, 20, and the two markers. The results on mar2 are shown as a three-color image (left), as well as separately for each of the probes (right).

# Genes involved and Proteins

## *SS18L1*

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

**DNA/RNA**

Genomic (chr20:60,169,869-60,189,352). Transcript of 4566 bp (NM_198935).

## *SSX1*

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

**DNA/RNA**

Genomic (chrX:47,999,741-48,011,823). Transcript of 1271 bp (NM_005635).

**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

Storlazzi CT, Mertens F, Mandahl N, Gisselsson D, Isaksson M, Gustafson P, Domanski HA, Panagopoulos I. A novel fusion gene, SS18L1/SSX1, in synovial sarcoma. Genes Chromosomes Cancer 2003;37:195-200.

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