

# SYT-SSX t(X;18) Translocations by RT-PCR

*USED TO DETECT THE PRESENCE OF t(X;18) TRANSCRIPTS IN TUMOR TISSUE WHERE SYNOVIAL SARCOMA IS SUSPECTED*

## Introduction

Synovial sarcomas account for 8–10 percent of all soft tissue sarcomas, with the t(X;18) (p11.2;q11.2) translocation serving as a specific marker for the disease. The *SYT* gene, a transcriptional co-activator, appears on chromosome 18 and is fused to a member of the *SSX* gene family. The *SSX* gene family contains nine members and is located on the X chromosome. Fusion to the *SSX1* gene accounts for the majority of synovial sarcomas, approximately 64 percent. The *SSX2* gene is fused to *SYT* in the vast remainder of cases, approximately 35 percent. Fusion of *SYT* to other members of the *SSX* family is rare. The type of *SYT-SSX* fusion transcript tends to associate with particular histological subtypes of synovial sarcoma. For example, the *SYT-SSX1* translocation is more common in the biphasic tumor type and the *SYT-SSX2* translocation is found more often in the monophasic tumor type.

## Clinical Background

The detection of *SYT-SSX* transcript is specific for synovial sarcoma and helps differentiate this tumor type from other neoplasms displaying an epithelioid, spindle cell, or combined morphology pattern. The prognostic significance of the different *SYT-SSX* transcripts is still unknown. Studies suggest that individuals with the *SYT-SSX1* translocation have a worse prognosis than those with the *SYT-SSX2* translocation.

## Interpretation

The t(X;18) results from an acquired translocation of the *SYT* gene to one of the *SSX* gene family members. Oligonucleotide primers are specific for the translocations, *SYT-SSX1* and *SYT-SSX2*. These translocations are detected by the presence of an amplification curve and an appropriate post-amplification melting temperature. A negative result does not exclude the presence of a t(X;18) chromosomal translocation to a different *SSX* family member.

## Methodology

Patient tumor RNA is isolated from formalin-fixed, paraffin-embedded tissue blocks and reverse transcribed into cDNA. The cDNA is then subjected to PCR amplification using oligonucleotide primers specific for the *SYT* gene on chromosome 18 and for the *SSX1* or the *SSX2* genes on the X chromosome. Each sample is also amplified for the *MRPL19* gene, which serves as a PCR control.

## Indications for Ordering

The principal use for this assay is to detect the presence of a *SYT-SSX*t(X;18) fusion transcript, which is specific for synovial sarcoma. Results of this test must always be interpreted in the context of histology and other relevant data, and should not be used alone for a diagnosis of malignancy. As currently configured, this assay is intended to serve as a qualitative test and should not be used to detect minimal residual disease.

## References

1. Kawai A, et al. *SYT-SSX* fusion as a determinant of morphology and prognosis in synovial sarcoma. *NEJM* 2003; 338:153–160.
2. Ladanyi M, et al. Impact of *SYT-SSX* fusion type on the clinical behavior of synovial sarcoma: a multi-institutional retrospective study of 243 patients. *Cancer Research* 2002; 62:135–140.
3. Tornkvist M, et al. A novel type of *SYT/SSX* fusion: methodological and biological implications. *Mod Pathology* 2002; 15:679–685.

## Test Information

**0040114**      ***SYT-SSX* t(X;18) Translocations by RT-PCR**

For specific collection, transport, and testing information, refer to the ARUP Web site at [www.aruplab.com](http://www.aruplab.com).