

# SS18 (SYT) (18q11) Gene Rearrangement by FISH

## Indications for Ordering

Diagnosis of synovial sarcoma in conjunction with histologic and clinical information

## Test Description

Fluorescence in situ hybridization

## Tests to Consider

### Primary test

[SS18 \(SYT\) \(18q11\) Gene Rearrangement by FISH 3001303](#)

- Molecular diagnosis of synovial sarcoma

### Related test

[Chromosome FISH, Interphase 2002298](#)

- Specific probe for *SS18 (SYT)* rearrangement must be requested
- Fresh tissue specimens only

## Disease Overview

### Incidence – rare

- Synovial sarcomas account for 8-10% of all soft tissue sarcomas

### Diagnostic/prognostic issues

- Synovial sarcomas may resemble other neoplasms, particularly those displaying an epithelioid, spindle cell, or combined morphology
- t(X;18)(p11.2;q11.2) translocation serves as a specific marker for synovial sarcoma
  - *SS18 (SYT)* gene fuses with *SSX* gene
    - Fusion with *SSX1* in ~65% of synovial sarcomas
    - Fusion with *SSX2* in ~35% of cases
    - Fusions with other *SSX* members (eg, *SSX4*) are rare
      - Specific fusion partners are not detected with this test
  - Fusion transcripts have associations with particular subtypes
    - *SS18-SSX1* is more common in biphasic tumor type
    - *SS18-SSX2* is more common in monophasic tumor type
- Prognosis
  - *SS18-SSX1* translocation may have a worse prognosis than *SS18-SSX2* translocation

## Genetics

**Translocations – *SS18-SSX1*, *SS18-SSX2***

### Structure/function

- *SS18* is located on chromosome 18
- *SSX1* and *SSX2* are located on the X-chromosome
- Each gene in the translocation codes for proteins that have opposite transcriptional functions
  - *SS18* – activator of oncogenes
  - *SSX1*, *SSX2* – tumor suppression

## Test Interpretation

### Results

- Positive – *SS18* rearrangement is detected
  - *SSX* translocation partner is not identified with this testing methodology
  - Synovial sarcoma likely
- Negative – no *SS18* rearrangement detected
  - Does not entirely exclude the presence of an *SS18* rearrangement as some translocations are cryptic and not evaluable by this testing methodology
  - Does not entirely exclude diagnosis of synovial sarcoma

### Limitations

- Testing using tissue fixed in alcohol-based or non-formalin fixatives has not been validated using this method
- *SS18* fusion partners are not detected with this test