

SS18 (SYT) (18q11) Gene Rearrangement by FISH in Synovial Sarcoma

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 2007222](#)

- 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-SSX2* translocation may have a worse prognosis than *SS18-SSX1* 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 a *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