

# SS18 (SYT) Gene Rearrangement by FISH

## FOR THE DIAGNOSIS OF SYNOVIAL SARCOMA

### Test Highlights

SS18 (SYT) FISH is a sensitive and specific method used to detect SYT gene rearrangements aiding in the diagnosis of synovial sarcoma.

#### Disease Overview

- Synovial sarcoma is a soft-tissue tumor that usually occurs near the joints in the legs and arms but has been documented in almost all tissue types.
- The separation of synovial sarcoma from other spindle-cell sarcomas and benign spindle-cell lesions is best achieved by identification of chromosome 18 (SYT) rearrangements.
- Since synovial sarcomas are high-grade neoplasms, identification of a spindle-cell sarcoma as a synovial sarcoma allows for the timely initiative of aggressive adjunct therapy.

#### Epidemiology

- Synovial sarcoma occurs primarily in the young, with a peak incidence before 30 years of age.
- Synovial sarcoma accounts for 5 to 10 percent of soft tissue sarcomas, with approximately 800 new cases diagnosed each year.

#### Genetics

Synovial sarcoma is strongly associated with t(X;18). This translocation involves the SYT gene on chromosome 18 and one of the three SSX genes on the X chromosome (SSX1, SSX2, and SSX4).

#### Pathophysiology

- The exact cause of synovial sarcoma is not known. Most patients with synovial sarcoma have an acquired t(X;18). These translocations can increase the rate of cell division and growth.
- The distinction of synovial sarcoma from other sarcomas is important and provides prognostic and therapeutically relevant information.

#### Indications for Ordering

Patients diagnosed with or suspected of having synovial sarcoma based on morphology or immunophenotypic studies.

#### Contraindications

This test is not recommended for detection of minimal residual disease.

#### Additional Ordering Notes

The biopsy site and fixative used should be provided. The submitted sample should contain sufficient viable tumor.

#### Interpretation

Presence of a SYT gene rearrangement is strongly supportive of a diagnosis of synovial sarcoma.

#### Limitations

- Tissues fixed in alcohol-based or non-formalin fixatives have not been tested using this method.
- This test will not identify the specific SS18 (SYT) translocation partner.

#### Methodology

- The detection of SYT gene rearrangements in formalin-fixed, paraffin-embedded tissue uses a commercially available DNA FISH probe.
- The presence of two fusion signals per nucleus indicates an intact SYT gene. The presence of a single orange and single green signal indicates a rearranged SYT gene.
- This test is conducted by noting the probe-signal configuration within the nuclei and reporting the result as rearranged or normal.

#### References

1. Bovee JV and Hogendoorn PC. Molecular pathology of sarcomas: concepts and clinical implications. *Virchows Arch* 2010; 456 (2):193–99.
2. Jain S, et al. Molecular classification of soft tissue sarcomas and its clinical applications. *Int J Clin Exp Pathol* 2010; 3(4):416–28.
3. de Alava E. Molecular pathology in sarcomas. *Clin Transl Oncol* 2007; 9(3) :130–44.
4. Sandberg AA, Bridge JA. Updates on the cytogenetics and molecular genetics of bone and soft tissue tumors synovial sarcoma. *Cancer Genet Cytogenet* 2002;133:1–23.

## Test Information

**0049380**

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

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

For information on test selection, ordering, and interpretation, refer to ARUP Consult® at [www.arupconsult.com](http://www.arupconsult.com).

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