Test Definition: SS18F

Synovial Sarcoma (SS), 18q11.2 (SS18 or SYT) Rearrangement, FISH, Tissue

Overview

Useful For

Identifying *SS18* gene rearrangements irrespective of the *SS18* fusion partner gene

Aiding in the diagnosis of synovial sarcoma by detecting a neoplastic clone associated with a gene rearrangement involving the *SS18 (SYT)* gene region at 18q11.2 when used conjunction with an anatomic pathology consultation

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_PBCT	Probe, +2	No, (Bill Only)	No
_PADD	Probe, +1	No, (Bill Only)	No
_PB02	Probe, +2	No, (Bill Only)	No
_PB03	Probe, +3	No, (Bill Only)	No
_IL25	Interphases, <25	No, (Bill Only)	No
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No

Testing Algorithm

This test includes a charge for probe application, analysis, and professional interpretation of results for one probe set (2 individual fluorescence in situ hybridization probes). Analysis charges are based on the number of cells analyzed per probe. No analysis charges apply if there are insufficient representative cells for evaluation.

Appropriate ancillary probes may be performed at the consultant's discretion to render comprehensive assessment. Any additional probes will have the results included within the final report and will be performed at an additional charge.

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type

Tissue

Ordering Guidance

This test does not include a pathology consultation. If a pathology consultation is requested, order PATHC / Pathology Consultation, and appropriate testing will be added at the discretion of the pathologist and performed at an additional charge.

Multiple oncology (cancer) gene panels are also available. For more information see Hematology and Oncology Test Selection Guide.

Additional Testing Requirements

Confirmation testing for the presence of a possible *SS18* fusion transcript by next-generation sequencing to resolve atypical or unbalanced fluorescence in situ hybridization results is available, order MCRSP / MayoComplete Targeted RNA Sequencing Panel, Next-Generation Sequencing, Tumor.

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

1. A pathology report is required for testing to be performed. If not provided, appropriate testing and/or interpretation may be compromised or delayed. Acceptable pathology reports include working drafts, preliminary pathology, or surgical pathology reports.

2. The following information must be included in the report provided:

-Patient name

-Block number - **must be on all blocks, slides, and paperwork**

-Collection date

-Tissue source

3. A reason for testing must be provided. If this information is not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.

Specimen Required

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Tissue block (fresh tissue is **not acceptable**)

Collection Instructions:

1. Submit a formalin-fixed, paraffin-embedded tumor tissue block.
2. Blocks prepared with alternative fixation methods (eg, Prefer, Bouin's) will be attempted but are less favorable for successful results. Provide fixation method used.

Additional Information:

1. Paraffin-embedded specimens can be from any anatomic location (skin, soft tissue, lymph node, etc).
2. Decalcified paraffin-embedded specimens will have testing attempted; however, the success rate is approximately 50%. **Testing may be canceled** if sufficient tumor tissue is not present.
3. **Submitted fresh tissue specimens will be canceled upon receipt**. If only fresh tissue is available, embed in paraffin prior to sending.

Acceptable:

Specimen Type: Tissue slides

Slides: 1 Hematoxylin and eosin-stained and 4 unstained
Collection Instructions: Submit 1 slide stained with hematoxylin and eosin and 4 consecutive unstained, positively charged, unbaked slides with 5-micron thick sections of the tumor tissue.
Additional Information: Broken or cracked unstained slides will be rejected. Stained slides that were broken and repaired (ie, double mounted) at the client site will not be rejected, as, in most cases, they can still be reviewed under the microscope. However, due to the double mounting, digital scanning cannot be performed.

Forms

If not ordering electronically, complete, print, and send Oncology Test Request (T729) with the specimen.

Specimen Minimum Volume

Slides: 1 Hematoxylin and eosin-stained and 2 unstained

Reject Due To

Broken or cracked unstained slides	Reject
Broken or cracked stained slides that cannot be repaired	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Tissue	Ambient (preferred)		
	Refrigerated		

Clinical & Interpretive

Clinical Information

Synovial sarcoma (SS) is a malignant soft tissue tumor that predominantly occurs in the lower limbs of children and young adults. This tumor accounts for approximately 5% to 10% of soft tissue tumors, has a poor prognosis, and may occur in other areas of the body such as the head and neck, heart, abdominal wall, mediastinum, and lung, in addition to the extremities. Accurate diagnosis of SS is important for appropriate clinical management of patients. Although immunohistochemical markers can be helpful in the correct diagnosis of these various tumor types, recent molecular studies have shown the superior specificity of molecular makers in differentiating SS from other tumors.

A recurrent, tumor-specific translocation t(X;18)(p11.2;q11.2) is observed in approximately 90% of synovial sarcomas. A single gene, *SS18 (SYT)*, has been implicated on 18q11.2, while 1 of 3 related genes, *SSX1, SSX2,* or infrequently *SSX4*, is

usually involved on Xp11.2. FISH testing can be used to identify *SS18* gene rearrangements in these tumors, which supports the diagnosis of SS.

Reference Values

An interpretive report will be provided.

Interpretation

SS18 will be clinically interpreted as positive, negative, or equivocal.

A neoplastic clone is detected when the percent of cells with an abnormality exceeds the normal cutoff for the SS18 probe set.

A positive result is consistent with rearrangement of the *SS18* gene and likely reflects *SS18* fusion with a partner gene. A positive result is consistent with a subset of synovial sarcoma. The significance of this FISH result is dependent on clinical and pathologic features.

A negative result suggests no rearrangement of the *SS18* gene region is present but does not exclude the diagnosis of synovial sarcoma. Clinical and pathologic correlation is required.

A negative result does not exclude the presence of a neoplastic disorder.

Cautions

This test is not approved by the US Food and Drug Administration, and it is best used as an adjunct to existing clinical and pathologic information.

This test is intended to be used for diagnostic purposes in synovial sarcoma.

This fluorescence in situ hybridization (FISH) assay does not rule out other chromosome abnormalities.

Fixatives other than formalin (eg, Prefer, Bouin's) may not be successful for FISH assays. Non-formalin fixed specimens will not be rejected.

Paraffin-embedded tissues that have been decalcified may not be successful for FISH analysis. The success rate of FISH studies on decalcified tissue is approximately 50%, but FISH will be attempted if sufficient tumor is present for analysis.

Fluorescence in situ hybridization studies will be attempted if sufficient tumor is present for analysis. The pathologist reviewing the hematoxylin and eosin-stained slide may find it necessary to cancel testing if insufficient tissue/tumor is available for testing.

If no FISH signal or a lack of sufficient tumor tissue is observed post-hybridization, the case will be released indicating a lack of FISH results.

Clinical Reference

1. WHO Classification of Tumours Editorial Board. Soft Tissue and Bone. 5th ed. IARC; 2020. World Health Organization Classification of Tumours. Vol 3

2. 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):1-23
3. Kokovic I, Bracko M, Golouh R, et al. Are there geographical differences in the frequency of SYT-SSX1 and SYT-SSX2 chimeric transcripts in synovial sarcoma?. Cancer Detect Prev. 2004;28(4):294-301
4. dos Santos NR, de Bruijn DR, van Kessel AG. Molecular mechanisms underlying human synovial sarcoma development. Genes Chromosomes Cancer. 2001;30(1):1-14

Performance

Method Description
The test is performed using a commercially available SS18 dual-color, break-apart strategy fluorescence in situ hybridization probe set. Paraffin-embedded tissue samples are cut at 5 microns and mounted on positively charged glass slides. The selection of tissue and the identification of target areas on the hematoxylin and eosin (H and E)-stained slide are performed by a pathologist. Using the H and E-stained slide as a reference, target areas are etched with a diamond-tipped engraving tool on the back of the unstained slide to be assayed. The probe set is hybridized to the appropriate target areas, and 2 technologists each analyze 50 interphase nuclei (100 total) with the results expressed as the percent of abnormal nuclei.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
7 to 10 days

Specimen Retention Time
Slides and H and E used for analysis are retained by the laboratory in accordance with regulatory requirements. Client provided paraffin blocks and extra unstained slides will be returned after testing is complete.

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

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Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

88271x2, 88291-DNA probe, each (first probe set), Interpretation and report
88271x2-DNA probe, each; each additional probe set (if appropriate)
88271x1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
88271x2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
88271x3-DNA probe, each; coverage for sets containing 5 probes (if appropriate)
88274 w/modifier 52-Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
88274-Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
SS18F	SS18, Synovial Sarcoma, FISH, Ts	93810-0

Result ID	Test Result Name	Result LOINC® Value
52131	Result Summary	50397-9
52133	Interpretation	69965-2
54582	Result	62356-1
CG742	Reason for Referral	42349-1
52134	Specimen	31208-2
52135	Source	31208-2
52136	Tissue ID	80398-1
52137	Method	85069-3
55024	Additional Information	48767-8
52138	Released By	18771-6
53833	Disclaimer	62364-5