

ETV6 (12p13.2) Rearrangement, FISH, Tissue

## Overview

## **Useful For**

Detection of ETV6 rearrangements irrespective of the ETV6 fusion partner gene

Supporting the diagnosis of many neoplasms including, but not limited to, mammary analogue secretory carcinoma, secretory carcinoma of the breast, and infantile fibrosarcoma when used in conjunction with pathologic assessment

#### **Reflex Tests**

Test Id	Reporting Name	Available Separately	Always Performed
_PBCT	Probe, +2	No	No
_PADD	Probe, +1	No	No
_PB02	Probe, +2	No	No
_PB03	Probe, +3	No	No
_IL25	Interphases, <25	No	No
_1099	Interphases, 25-99	No	No
_1300	Interphases, >=100	No	No

### **Testing Algorithm**

This test includes a charge for the probe application, analysis, and professional interpretation of results for one probe set (2 individual fluorescence in situ hybridization probes). No analysis charges will be incurred if an insufficient number of representative cells are available for analysis.

Appropriate ancillary probes may be performed at consultant 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



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charge.

Multiple oncology (cancer) gene panels are also available. For more information see <u>Hematology, Oncology, and Hereditary Test Selection Guide</u>

## **Additional Testing Requirements**

Confirmation testing by next-generation sequencing to resolve atypical or unbalanced fluorescence in situ hybridization results of this gene region is available, order SARCP / Sarcoma Targeted Gene Fusion/Rearrangement 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
- -Date of collection
- -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

**Collection Instructions:** Submit a formalin-fixed, paraffin-embedded tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used.

#### **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.

## **Forms**

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

## **Specimen Minimum Volume**

Slides: 1 Hematoxylin and eosin stained and 2 unstained

# Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

# **Specimen Stability Information**



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

# Clinical & Interpretive

#### **Clinical Information**

ETV6 rearrangement has been identified in a wide variety of neoplasms including mammary analogue secretory carcinoma, secretory carcinoma of the breast, and infantile fibrosarcoma.

# **Reference Values**

An interpretive report will be provided.

## Interpretation

A neoplastic clone is detected when the percent of cells with an abnormality exceeds the normal cutoff for the ETV6 fluorescence in situ hybridization (FISH) probe set.

A positive result is consistent with the presence of *ETV6* rearrangement and likely reflects *ETV6* fusion with a partner gene. The significance of this FISH result is dependent on clinical and pathologic features.

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 is best used as an adjunct to existing clinical and pathologic information.

Fixatives other than formalin (eg, Prefer, Bouin's) may not be successful for fluorescence in situ hybridization (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.

If no FISH signals are observed post-hybridization, the case will be released indicating a lack of FISH results.

# **Clinical Reference**

- 1. WHO Classification of Tumours Editorial Board. Breast Tumours. 5th ed. IARC; 2019. WHO Classification of Tumours. Vol 2
- 2. WHO Classification of Tumours Editorial Board. Soft Tissue and Bone Tumours. 5th ed. IARC; 2020. WHO Classification of Tumours. Vol 3
- 3. Skalova A, Vanecek T, Martinek P, et al. Molecular profiling of mammary analog secretory carcinoma revealed a subset of tumors harboring a novel ETV6-RET translocation. Am J Surg Pathol. 2018;42(2):234-246
- 4. Skalova A. Mammary analogue secretory carcinoma of salivary gland origin: an update and expanded morphologic and immunohistochemical spectrum of recently described entity. Head Neck Pathol 2013;7:S30-S36
- 5. Makretsov N, He M, Hayes M, et al. A fluorescence in situ hybridization study of ETV6-NTRK3 fusion gene in secretory breast carcinoma. Genes Chromosomes Cancer. 2004;40(2):152-157
- 6. Sheng WQ, Hisaoka M, Okamoto S, et al. Congenital-infantile fibrosarcoma. A clinicopathologic study of 10 cases and



ETV6 (12p13.2) Rearrangement, FISH, Tissue

molecular detection pf the ETV6-NTRK3 fusion transcripts using paraffin-embedded tissues. Am J Clin Pathol. 2001;115(3):348-355

- 7. Steelman C, Katzenstein H, Parham D, et al. Unusual presentation of congenital infantile fibrosarcoma in seven infants with molecular-genetic analysis. Fetal Pediatr Pathol. 2011;30(5):329-337
- 8. Skalova A, Vanecek T, Sima R, et al. Mammary analogue secretory carcinoma of salivary glands, containing the ETV6-NTRK3 fusion gene: a hitherto undescribed salivary gland tumor entity. Am J Surg Pathol. 2010;34(5):599-608

#### **Performance**

## **Method Description**

The test is performed using a laboratory-developed ETV6 (12p13.2) dual-color, break-apart strategy probe (BAP). Formalin-fixed, paraffin-embedded tissues 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 independently 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 <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

## **Test Classification**



ETV6 (12p13.2) Rearrangement, FISH, Tissue

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)

88275 -Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

### **LOINC®** Information

Test ID	Test Order Name	Order LOINC® Value
ETV6F	ETV6 (12p13.2), FISH, Ts	101380-4

Result ID	Test Result Name	Result LOINC® Value
46837	Result Summary	50397-9
46838	Interpretation	69047-9
46839	Result	62356-1
46840	Reason For Referral	42349-1
46841	Specimen	31208-2
46842	Source	39111-0
46843	Tissue ID	80398-1
46844	Method	85069-3
46845	Additional Information	48767-8
46846	Disclaimer	62364-5
46847	Released By	18771-6