MAYO CLINIC LABORATORIES

Test Definition: FGFR2

FGFR2 (10q26.1) Rearrangement, FISH, Tissue

Overview

Useful For

Detection of *FGFR2* rearrangements irrespective of the *FGFR2* fusion partner gene

Providing prognostic information and guiding treatment for patients with cholangiocarcinomas and other tumor types including bladder, thyroid, oral cavity, and brain

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 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

charge.

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

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 (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.

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

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

Clinical & Interpretive

Clinical Information

Cholangiocarcinoma is a malignancy arising from the biliary tract epithelium. These tumors are often clinically advanced at the time of presentation, and the prognosis is very poor with a short overall survival. Treatment is generally limited to surgical resection, which is associated with a high degree of morbidity, and palliative chemotherapy regimens. Therefore, additional treatment options are eagerly sought.

Rearrangement of the *FGFR2* gene region has been identified in a subset of cholangiocarcinomas. These rearrangements result in overexpression of *FGFR2*, which offers the possibility of using targeted *FGFR2*-inhibitor therapy for treatment. *FGFR2* rearrangements have also been identified in a number of other cancers including bladder, thyroid, oral cavity, and brain.

Reference Values

An interpretive report will be provided.

Interpretation

FGFR2 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 FGFR2 fluorescence in situ hybridization (FISH) probe set.

A positive result is consistent with the presence of an *FGFR2* rearrangement and likely reflects *FGFR2* fusion with a partner gene. The significance of this FISH result is dependent on additional clinical and pathologic features.

A negative result suggests no rearrangement of the *FGFR2* gene region is present.

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

Cautions

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

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 signals or a lack of sufficient tumor tissue are observed post-hybridization, the case will be released indicating a lack of FISH results.

Clinical Reference

1. Mitesh BJ, Champion MD, Egan JB, et al. Integrated Genomic Characterization Reveals Novel, Therapeutically Relevant Drug Targets in FGFR and EGFR Pathways in Sporadic Intrahepatic Cholangiocarcinoma. PLOS Genetics. 2014;10(2):e1004135
2. Graham RP, Barr Fritcher EG, Pestova E, et al. Fibroblast growth factor receptor 2 translocations in intrahepatic cholangiocarcinoma. Hum Pathol. 2014;45(8):1630-1638
3. Arai Y, Totoki Y, Hosoda F, et al. Fibroblast growth factor receptor 2 tyrosine kinase fusions define a unique molecular subtype of cholangiocarcinoma. Hepatology. 2014;59(4):1427-1434
4. Wu YM, Su F, Kalyana-Sundaram S, et al. Identification of targetable FGFR gene fusions in diverse cancers. Cancer Discov. 2013;3(6):636-647
5. Zou Y, Zhu K, Pang Y, et al. Molecular detection of FGFR2 rearrangements in resected intrahepatic cholangiocarcinomas: FISH could be an ideal method in patients with histological small duct subtype. J Clin Transl Hepatol. 2023;11(6):1355-1367

Performance

Method Description

The test is performed using a laboratory-developed FGFR2 (10q26.13) dual-color, break-apart strategy fluorescence in situ hybridization (FISH) probe set (BAP). 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 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 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.

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

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
FGFR2	FGFR2 (10q26.1), FISH, Ts	95784-5

Result ID	Test Result Name	Result LOINC® Value
38094	Result Summary	50397-9
38095	Interpretation	69965-2
38096	Result	62356-1
38097	Reason For Referral	42349-1
38098	Specimen	31208-2
38099	Source	31208-2

38100	Tissue ID	80398-1
38101	Method	85069-3
38102	Additional Information	48767-8
38103	Disclaimer	62364-5
38104	Released By	18771-6