Test Definition: FGFRF  
FGFR1 (8p11.2), FISH

Overview

Useful For
An aid in identifying patients with myeloproliferative syndromes and the t(8;var)(p11.2;var) translocation who therefore are likely resistant to current chemotherapies

Reflex Tests

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Testing Algorithm
This test includes a charge for application of the first probe set (2 FISH probes) and professional interpretation of results. Additional charges will be incurred for all reflex probes performed. Analysis charges will be incurred based on the number of cells analyzed per probe set. If no cells are available for analysis, no analysis charges will be incurred.

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Varies

Specimen Required
Provide a reason for referral with each specimen. The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.

Advise Express Mail or equivalent if not on courier service.

Submit only 1 of the following specimens:

Specimen Type: Blood

Container/Tube: Green top (sodium heparin)
Specimen Volume: 7-10 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.

Specimen Type: Bone marrow

Container/Tube: Green top (sodium heparin)

Specimen Volume: 1-2 mL

Collection Instructions:
1. Invert several times to mix bone marrow.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.

Forms
If not ordering electronically, complete, print, and send a Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Minimum Volume
Blood: 2 mL
Bone Marrow: 1 mL

Reject Due To
No specimen should be rejected.

Specimen Stability Information

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Clinical and Interpretive

Clinical Information
The gene for fibroblast growth factor receptor 1 (FGFR1) is located at 8p11.2 and rearrangements of FGFR1 are found in stem cell myeloproliferative disorders involving both lymphoid and myeloid lineages. The stem cell myeloproliferative disorders with FGFR1 rearrangements are also called 8p11 (eight p11) myeloproliferative syndromes (EMS) and have variable presentations. EMS often transform rapidly into myelomonocytic leukemia and generally have a poor outcome due to resistance to current chemotherapies, including imatinib mesylate; median survival is about 12 months.
All translocations affecting FGFR1 have a similar structure with a 5’ gene partner translocating to the 3’ FGFR1 at exon 9. The fusion transcripts encode large proteins containing the N-terminus of the translocation partner, and the tyrosine kinase domain of FGFR1 in the C-terminus. Leukemogenesis is caused by inappropriate activation of FGFR1.

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 any given probe.

The presence of a positive clone supports a diagnosis of malignancy.

The absence of an abnormal clone does not rule out the presence of 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.

Bone marrow is the preferred specimen type for this FISH test. If bone marrow is not available, a blood specimen may be used if there are malignant cells in the blood specimen (as verified by hematopathology).

Supportive Data
A blinded study using the FGFR1 dual-color break-apart (BAP) probe set was performed on 44 blood and bone marrow fixed cell pellets from 19 patients with t(8p11.2;var) identified by chromosome analysis and 25 normal control specimens. Rearrangement of FGFR1 was identified in 17 of 19 neoplastic specimens. The FGFR1 gene rearrangement was not detected in any of the 25 control specimens. The normal controls were used to generate a normal cutoff for this assay.

Clinical Reference


3. WHO Classification of Tumours of Hematopoietic and Lymphoid Tissues. Edited by SH Swerdlow, E Campo, NL Harris, et al. Published by the International Agency for Research on Cancer (IARC), 150 cours Albert Thomas, 69372 Lyon Cedex 08, France, 2008, pp 72-73

Performance

Method Description
The test uses a laboratory-developed FGFR1 dual-color break-apart (BAP) strategy probe. The probe set is hybridized to the sample and 2 technologists each analyze 100 interphase nuclei (200 total) with the results expressed as the percent abnormal nuclei. (Unpublished Mayo method)

PDF Report
No
Day(s) and Time(s) Test Performed
Samples processed Monday through Sunday. Results reported Monday through Friday, 8 a.m.-5 p.m. CST.

Analytic Time
7 days

Maximum Laboratory Time
10 days

Specimen Retention Time
Three weeks.

Performing Laboratory Location
Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information
88271x2, 88291 â€“ DNA probe, each (first probe set), Interpretation and report

88271x2 â€“ DNA probe, each; each additional probe set (if appropriate)

88271x1 â€“ DNA probe, each; coverage for sets containing 3 probes (if appropriate)

88271x2 â€“ DNA probe, each; coverage for sets containing 4 probes (if appropriate)

88271x3 â€“ DNA probe, each; coverage for sets containing 5 probes (if appropriate)

88274 w/modifier 52 â€“ Interphase in situ hybridization, <25 cells, each probe set (if appropriate)

88274 â€“ Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)

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

LOINC® Information

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