

Overview

Useful For

Identifying patients with chronic myelomonocytic leukemia and other hematologic disorders who may be responsive to imatinib mesylate

Identifying and tracking chromosome abnormalities and response to therapy

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,	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 application of the first probe set (2 FISH probes) and professional interpretation of results. Additional charges will be incurred for application of 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

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

Specimen Stability Information

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

Clinical and Interpretive

Clinical Information

Platelet-derived growth factor receptor-beta (PDGFRB) produces a tyrosine kinase involved in cell proliferation. Translocation-ets-leukemia protein (encoded by the gene *ETV6*) is a gene transcription protein that is frequently rearranged in leukemias. A 5;12 translocation, t(5;12)(q33;p13), results in a fusion product (*PDGFRB/ETV6*) that is

seen in approximately 1% to 2% of patients diagnosed with chronic myelomonocytic leukemia. Patients with this translocation often have associated hypereosinophilia.

Imatinib mesylate is an inhibitor of tyrosine kinases, including PDGFRB. Patients with the 5;12 translocation are reportedly responsive to imatinib mesylate; upon treatment, they usually go into complete remission.

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.

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.

Supportive Data

A blinded study using the *PDGFRB/ETV6* dual-color, dual-fusion (D-FISH) strategy probe was performed on 12 samples from patients identified with a t(5;12) by chromosome analysis and a series of normal control specimens. Translocation of *PDGFRB* and *ETV6* was identified in the neoplastic specimens but was not detected in any of the control specimens. The normal controls were used to generate a normal cutoff for this assay.

Clinical Reference

1. Pardanani A, Reeder T, Porrata LF, et al: Imatinib therapy for hypereosinophilic syndrome and other eosinophilic disorders. *Blood* 2003;101:3391-3397
2. Cain JA, Grisolan JL, Laird AD, et al: Complete remission of TEL-PDGFRB-induced myeloproliferative disease in mice by receptor tyrosine kinase inhibitor SU11657. *Blood* 2004;104:561-564

Performance

Method Description

This test is performed using a laboratory developed *PDGFRB/ETV6* dual-color, dual-fusion (D-FISH) strategy probe. The probe set is hybridized to the sample and 2 technologists each analyze 250 interphase nuclei (500 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. to 5 p.m. CST.

Analytic Time

5 days

Maximum Laboratory Time

7 days

Specimen Retention Time

4 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 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
512F	PDGFRB/TEL, FISH	21816-4

Result ID	Test Result Name	Result LOINC Value
51863	Result Summary	50397-9
51865	Interpretation	78227-6
51864	Result Table	93356-4
54540	Result	62356-1
CG673	Reason for Referral	42349-1
CG674	Specimen	31208-2



Result ID	Test Result Name	Result LOINC Value
51866	Source	31208-2
51867	Method	49549-9
54452	Additional Information	30954-2
53873	Disclaimer	62364-5
51868	Released By	18771-6