

Overview

Useful For

Providing diagnostic information and helping to determine whether a targeted *JAK2* inhibitor could be useful for therapy

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,	No	No
_I099	Interphases, 25-99	No	No
_I300	Interphases, >=100	No	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

Necessary Information

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.

Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Blood

Container/Tube: Green top (sodium heparin)

Specimen Volume: 4 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

The *JAK2* gene is a protein tyrosine kinase involved in cytokine signaling. Chromosomal translocations involving *JAK2* can lead to the formation of chimeric oncoproteins in hematologic malignancies. Rearrangements involving 9p24.1 are typically aggressive and rare abnormalities seen in various hematologic diseases. *JAK2* inhibitors are one of the only therapy options besides a stem cell transplant for *JAK2* rearrangements.

Reference Values

An interpretive report will be provided.

Interpretation

A positive result is detected when the percent of cells with an abnormality exceeds the normal cutoff for the probe set.

A positive result suggests rearrangement of the *JAK2* locus. A negative result suggests no rearrangement of the *JAK2* gene region at 9p24.1.

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.

Supportive Data

FISH analysis was performed on 2 bone marrow samples from patients with previously identified 9p24.1 abnormality and 25 noncancerous blood and bone marrow control specimens. Rearrangement of *JAK2* was identified in 2 samples. The normal controls were used to generate a normal cutoff for this assay.

Clinical Reference

1. Chase A, Bryant C, Score J, et al: Ruxolitinib as potential targeted therapy for patients with JAK2 rearrangements. *Haematol* 2013;98(3):404-408
2. Van Roosbroeck K, Cox L, Tousseyn T, et al: *JAK2* rearrangements, including the novel *SEC31A-JAK2* fusion, are recurrent in classical Hodgkin lymphoma. *Blood* 2011;117(15):4056-4064
3. Roberts K, Li Y, Payne-Turner D, et al: Targetable Kinase-Activating Lesions in Ph-like Acute Lymphoblastic Leukemia. *N Engl J Med* 2014;371:1005-101

Performance

Method Description

The test is performed using a laboratory-developed *JAK2* (9p24.1) dual-color break-apart strategy probe (BAP). The probe set is hybridized to the appropriate target areas and 2 technologists each analyze 100 interphase nuclei (200 total) with the results expressed as the percent of abnormal nuclei.(Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday through Friday, 8 a.m.-5 p.m.

Analytic Time

7 days

Maximum Laboratory Time

10 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

88271 x 2, 88291-DNA probe, each (first probe set), Interpretation and report

88271 x 2-DNA probe, each; each additional probe set (if appropriate)

88271 x 1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)

88271 x 2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)

88271 x 3-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
JAK2F	JAK2 (9p24.1) Rearrangement, FISH	In Process

Result ID	Test Result Name	Result LOINC Value
43674	Result Summary	50397-9
43675	Interpretation	69965-2
43676	Result Table	93356-4
43677	Result	62356-1
CG972	Reason for Referral	42349-1
CG973	Specimen	31208-2
43680	Source	31208-2
43681	Method	49549-9
43682	Additional Information	48767-8
43683	Disclaimer	62364-5
43684	Released By	18771-6

