MAYO CLINIC LABORATORIES

Test Definition: JAK2P

JAK2 (9p24.1) Rearrangement, Hematologic Disorders, FISH, Tissue

Overview

Useful For

Providing diagnostic information for hematologic malignancies

Aiding in the determination of 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, <25	No	No
_I099	Interphases, 25-99	No	No
_I300	Interphases, >=100	No	No

Testing Algorithm

This test does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered, and the appropriate fluorescence in situ hybridization (FISH) test will be performed at an additional charge.

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.

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.

Special Instructions

- Anaplastic Large Cell Lymphoma Evaluation Algorithm

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type
Tissue

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information
Provide a reason for testing and pathology report 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: Tissue
Preferred: Tissue block
Collection Instructions: Submit a formalin-fixed, paraffin-embedded (FFPE) tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used.

Acceptable: Slides
Collection Instructions: Four consecutive, unstained, 5 micron-thick sections placed on positively charged slides, and 1 hematoxylin and eosin-stained slide.

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

Specimen Minimum Volume
Two consecutive, unstained, 5- micron- thick sections placed on positively charged slides, and 1 hematoxylin and eosin-stained slide.

Reject Due To
No specimen should be rejected.

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

The *JAK2* gene codes for 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 rare abnormalities seen in various hematologic diseases and are typically aggressive. Identification of opportunities to apply targeted therapy with *JAK2* inhibitors can be helpful for patients with *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.

Fixatives other than formalin (eg, Prefer, Bouin) may not be successful for fluorescence in situ hybridization (FISH) assays, however nonformalin-fixed samples will not be rejected.

Paraffin-embedded tissues that have been decalcified are generally unsuccessful for FISH analysis. The pathologist reviewing the hematoxylin and eosin-stained slide may find it necessary to cancel testing.

Supportive Data

Fluorescence in situ hybridization (FISH) analysis was performed on 2 paraffin-embedded tissue samples from patients with previously identified 9p24.1 abnormality and 25 noncancerous lymph node 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. Haematologica. 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
4. Springuel L, Renauld JC, Knoops L: JAK kinase targeting in hematologic malignancies: a sinuous pathway from identification of genetic alteration towards clinical indications. Haematologica. 2015;100:1240-1253
5. Reshmi SC, Harvey RC, Roberts KG, et al: Targetable kinase gene fusions in high-risk B-ALL: A study from the Children's Oncology Group. Blood. 2017;129:3352-3361

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 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: Indefinitely. Client provided paraffin blocks and extra unstained slides (if provided) will be returned after testing is complete.

Performing Laboratory Location
Rochester

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
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

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Test ID	Test Order Name	Order LOINC® Value
JAK2P	JAK2 (9p24.1) Rearrangement,FISH,Ts	In Process

Result ID	Test Result Name	Result LOINC® Value
606822	Result Summary	62357-9
606824	Result	62356-1
GC052	Reason for Referral	42349-1
606823	Interpretation	69965-2
606825	Specimen	31208-2
606826	Source	31208-2
606827	Tissue ID	80398-1
606828	Method	85069-3
606829	Additional Information	48767-8
606830	Disclaimer	62364-5
606831	Released By	18771-6