

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**

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**

- 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 Definition: JAK2P

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

| Test ID | Test Order Name                        | Order LOINC® Value |
|---------|--|--------------------|
| JAK2P   | JAK2 (9p24.1)<br>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             |