



PAML Offers Real-Time PCR for the JAK2 (V617F) Mutation

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PAML is pleased to offer real-time PCR testing for the JAK2 (V617F) mutation associated with myeloproliferative disorders.

Myeloproliferative disorders are clonal hematologic malignant diseases including chronic myelogenous leukemia (CML), polycythemia vera (PV), idiopathic myelofibrosis (IMF), and essential thrombocytosis (ET), among other disorders. Frequently, diagnosis is simple, with clinical findings allowing definitive determination. In the case of CML, a specific chromosomal abnormality, the Philadelphia chromosome [t(9;22)(q34;q11)], defines the disease. In other cases, however, it may be difficult to differentiate reactive bone marrow findings from chronic infection, infiltration (in the case of ET and IMF), or secondary responses (such as differentiating hypoxia-induced erythrocytosis from PV). Recently a high-frequency mutation in the gene for the Janus kinase 2 (JAK2) signaling protein was described in patients with PV, ET, and IMF. Identification of this mutation in patients may allow differentiation of myeloproliferative syndromes from reactive disorders.

The JAK2 protein is a tyrosine kinase involved in the down-regulation of a signal transduction pathway in hematopoietic cells. A single base mutation in the *JAK2* gene, located on chromosome 9 (9p24.1), results in a phenylalanine for valine substitution (V617F) in the JH2 kinase domain of the protein, resulting in constitutive activation of the protein kinase and red blood cell and platelet proliferation. This mutation is commonly found in patients with PV, ET, and MF, and less frequently in myelodysplasia and chronic myelomonocytic leukemia (3-5%).

Test sensitivity allows detection of a 1% cell population harboring the JAK2 V617F mutation.

References

James et al. Nature 434: 1144-48, 2005

Baxter et al. The Lancet 365: 1054-61, 2005

Karlovics et al. NEJM 353: 1779-90, 2005

Quick Facts

Frequency of JAK2 V617F

- ▶ Polycythemia vera 74-97%
- ▶ Essential thrombocytosis 33-57%
- ▶ Idiopathic myelofibrosis 35-50%

May be useful to:

- ▶ Confirm myeloproliferative disorder
- ▶ Avoid erythroid colony assays
- ▶ Help predict response to JAK2 kinase inhibitors (may be clinically available soon)

For more information, please
contact Client Services or
see us on the Web at



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TRI-CITIES LABORATORY • TREASURE VALLEY LABORATORY • ALPHA MEDICAL LABORATORY

For more information, please contact your local representative.

Test Information

DESCRIPTION **JAK2 (V617F) MUTATION BY PCR**

METHOD PCR

ORDER CODE JAK2

CPT CODES 83891, 83896 × 2, 83898, 83903, 83912

SAMPLE 3 mL whole blood EDTA (lavender top tube) or 1 mL bone marrow EDTA (lavender top tube). Store and transport at room temperature. If delayed more than 72 hours, store and transport refrigerated.

COMMENTS *Minimum amount:* 1mL whole blood EDTA, 0.5 mL bone marrow EDTA

Unacceptable conditions: serum, heparinized whole blood or bone marrow, frozen whole blood or bone marrow, severely hemolyzed whole blood, specimens submitted in leaky containers or over 5 days old, specimens not received in original unopened tube.

Alternative samples: sodium citrate or ACD whole blood (blue or yellow top tube).

Stability: 72 hours at room temperature, 5 days refrigerated, unstable frozen.

SCHEDULE Weekly

TURNAROUND 2-5 days

RANGES Negative