



UW Medicine  
Department of Laboratory Medicine

## JAK2 V617F Mutation Test

[Laboratory Medicine Home Page](#)

### For Clinical Information:

Harvey Greisman, MD, PhD,  
Director of Hematology  
(206) 598-6131

### For Technical Information:

Harvey Greisman, MD, PhD,  
Director of Hematology  
(206) 598-6131

### University of Washington

### Department of Laboratory Medicine

*For additional ordering  
information and instructions,  
contact the Community Services  
office at (206) 598-6066 or  
(800) 713.5198*

We are pleased to announce, beginning September 1, 2005, a qualitative assay for the JAK2 V617F point mutation in patients with myeloproliferative diseases (MPD). Leukocytes from the majority of polycythemia vera (PV) patients and up to half of essential thrombocythemia (ET) patients and idiopathic myelofibrosis (IMF) patients have been found to carry this activating point mutation in the auto-inhibitory domain of the JAK2 tyrosine kinase. This test may obviate the need for endogenous erythroid colony (EEC) testing in MPD patients. The mutation also has been found in a minority of patients with other myeloid stem cell disorders, including chronic myelomonocytic leukemia (CMML) and myelodysplastic syndromes (MDS).

In our assay, genomic DNA is purified from patient leukocytes and amplified using two primers that flank JAK2 exon 12 and a third primer that hybridizes specifically to the mutant V617F allele. A control 364 nucleotide fragment is produced by the flanking primers regardless of mutation status and a unique 203 nucleotide fragment is produced by the allele-specific primer only if the V617F allele is present. Fluorescently-labeled primers are used so that the amplified products can be separated and detected by capillary electrophoresis. The mutation might not be detected (false negatives) in samples that contain too many non-myeloid cells (e.g. lymphocytes), which do not harbor the acquired JAK2 mutation, or in specimens containing too few granulocytes or monocytes.

- Specimen: 5-10 mL blood (EDTA/purple top) or 1-2 mL bone marrow (EDTA). Specimen must be labeled with two patient identifiers, i.e. name, date of birth, or patient ID number. (NOTE: JAK2 and BCR-ABL testing can be performed on a single specimen if both tests are requested at the same time. Because a separate DNA extraction step is required, the JAK2 test cannot be added after a specimen has been processed for BCR-ABL testing.)
- Packaging: Send specimens in a labeled, leak-proof tube. Wrap the tube in absorbent material and place in a crush-proof container. Complete a UW Hematopathology requisition and place in a separate plastic bag in the shipping container.
- Shipping: Send entire sample at room temperature or on ice (refrigeration is recommended if sample will not arrive at UW within 24-48 hours).
- Ship to: Hematopathology Laboratory UW Medical Center  
Room NW 225  
1959 NE Pacific Street  
Seattle, WA 98195
- CPT codes: 83891, 83894, 83896 (x2), 83898, 83912
- Test Order Code: JAK2