**JAK2 gene V617F quantitative**

**Indications for testing**
The JAK2 V617F mutation is the most prevalent mutation in BCR/ABL1 negative myeloproliferative neoplasms (MPNs). The JAK2 gene is located on chromosome 9p24 and expresses the Janus kinase 2 non-receptor tyrosine kinase. JAK2 V617F is a somatic G to T point mutation at position 1849 in exon 14 of JAK2 that is associated with uncontrolled blood cell growth. JAK2 V617F/G1849T mutations occur in ~ 95% of patients with polycythemia vera (PV), ~55% with essential thrombocythemia (ET) and ~65% with primary myelofibrosis (PMF).

**Testing Methodology**
The assay detects the JAK2 V617F/G1849T mutation using PCR primers and allele-specific dye-labeled oligonucleotides that exactly match the mutant or normal alleles. When perfectly hybridized to the template DNA sequence, the labeled oligonucleotides are cleaved by the 5' -> 3' exonuclease activity of DNA polymerase. Cleavage during the exponential phase of PCR releases the reporter dye from the quencher and the signal is quantified ("real time quantitative PCR"). The results report the mutant allele proportion of total JAK2 alleles.

The validated limit of detection for this assay is 0.1%.

**Specimen Requirements**
- **Peripheral blood**--1 lavender-top (EDTA) tube. Invert several times to mix blood. Forward promptly at ambient temperature.
- **Bone Marrow**--Place 1-2 mL of anticoagulated bone marrow in a lavender-top (EDTA) tube. Invert several times to mix bone marrow. Forward promptly at ambient temperature.

**Clinical information must be provided with specimen referral in order to correctly interpret test results.**

Molecular Diagnostic Laboratory  
Barnes-Jewish Hospital, Institute of Health  
Mail Stop 90-28-344  
425 South Euclid Avenue, Room 5970  
St. Louis, MO 63110

**Current Pricing**
Contact Lab Customer Service for current pricing 314 362-1470.  
CPT code: 81270