Molecular Diagnostic Laboratory
Phone: (314) 454-8685
FAX: (314) 454-7616

Jacqueline Payton, MD, PhD, FCAP
Medical Director
http://pathology.wustl.edu/patientcare/moldiagnostic.ph

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.

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

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

Current Pricing

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

1. Campbell PJ, Green AR. The myeloproliferative disorders. *N. Engl. J. Med.* 2006;355(23):2452-66.

2. Lippert E, Girodon F, Hammond E, et al. Concordance of assays designed for the quantification of JAK2V617F: a multicenter study. Haematologica 2009;94:38-45.

3. Jovanovic JV, Ivey A, Vannucchi AM, et al. Establishing optimal quantitative-polymerase chain reaction assays for routine diagnosis and tracking of minimal residual disease in JAK2-V617F-associated myeloproliferative neoplasms: a joint European LeukemiaNet/MPN&MPNr-EuroNet (COST action BM0902) study. Leukemia 2013;27:2032-9.

4. Skoda RC, Duek A, Grisouard J. Pathogenesis of myeloproliferative neoplasms. Experimental Hematology 2015; 43: 599-608