JAK2 V617F Quantitative Assay

Description:

JAK2 is a tyrosine kinase that is important in signal transduction in hematopoietic cells. A specific mutation in the *JAK2* gene, known as V617F, has been found to be present in cells from patients with chronic myeloproliferative disorders. V617F is an activating mutation in exon 14. This mutation can be helpful for clinicians in distinguishing between reactive cytosis and myeloproliferative disorders. Additionally, in the future, patients with this specific mutation might benefit from targeted therapies designed to inhibit the tyrosine kinase activity of JAK2.

Indications:

The main indication for *JAK2* testing at this time is diagnosis. The *JAK2* V617F mutation can help a clinician distinguish overlapping clinical phenotypes and make a diagnosis of a myeloproliferative disorder such as polycythemia vera, essential thrombocythemia, and/or myeloid metaplasia with myelofibrosis. In the future, JAK2 V617F testing might aid in providing information that can be used to aid in treatment related decisions or in prognostication.

Specimen:

Note: Sample should be received same day or shipped overnight on ice (do NOT freeze) within 48 hours of draw. 5-10mLs peripheral blood or 3-5mLs bone marrow in EDTA tube.

Methodology:

We use the JAK2 MutaQuant™ Assay from Ipsogen. This highly sensitive approach involves the use of a SNP specific forward primer resulting in the selective amplification of mutant or wild type allele. The fluorescent labeled products are easily detectable on a real-time PCR instrument. The quantities of normal and mutated *JAK2* copies are measured using a standard curve and the relative percentage of genes that possess this mutation detected (%V617) is reported.

Sensitivity:

The *JAK2* MutaQuant™ Assay is designed to detect the *JAK2* V617F mutation among at least 10,000 normal copies of the gene. The analytic sensitivity of this assay has been determined by this laboratory to be 0.10%. The variation of this assay, though, can be approximately 5% between laboratories and within a laboratory. We consider results above 0.10% to be positive for presence of the *JAK2* mutation.

Turn Around Time:

7-10 days.

Billing and CPT Codes:

JAK2 Quantitative (V617F): 81270
Please call 1-866-450-4198 for any pricing or billing questions.

Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Shipping Instructions:

Please enclose **test requisition** with sample. **All information must be completed before sample can be processed.** Place samples in Styrofoam mailer and ship on ice (do NOT freeze) by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Genetics and Genomics Diagnostic Laboratory
3333 Burnet Avenue NRB 1013
Cincinnati, OH 45229
513-636-4474

Cincinnati Children's®
changing the outcome together

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References:

Delhommeau, F., S. Dupont, et al. (2007) Evidence That the JAK2 G1849T (V617F) Mutation Occurs in a Lymphomyeloid Progenitor in Polycythemia Vera and Idiopathic Myelofibrosis. Blood 109(1): 71-7.

James, C., V. Ugo, et al. (2005) A Unique Clonal JAK2 Mutation Leading to Constitutive Signalling Causes Polycythaemia Vera. Nature 434(7037): 1144-8.

Kralovics, R., F. Passamonti, et al. (2005) A Gain-of-Function Mutation of JAK2 in Myeloproliferative Disorders. The New England Journal of Medicine 352(17): 1779-90.

Lacout, C., D.F. Pisani, et al. (2006) JAK2V617F Expression in Murine Hematopoietic Cells Leads to MPD Mimicking Human PV with Secondary Myelofibrosis. Blood 108(5): 1652-60.

Martinaud, C., P. Brisou, et al. (2010) Is the JAK2(V617F) Mutation Detectable in Healthy Volunteers?. American Journal of Hematology 85(4): 287-8.

Perricone et al. Oncotarget. 2017 Jun 6; 8(23): 37239–37249

Rapado, I., E. Albizua, et al. (2008) Validity Test Study of JAK2 V617F and Allele Burden Quantification in the Diagnosis of Myeloproliferative Diseases. Annals of Hematology 87(9): 741-9.

Rumi, E., D. Pietra, et al. (2014) Clinical Effect of Driver Mutations of JAK2, CALR, or MPL in Primary Myelofibrosis. Blood 124(7): 1062-9.

Sidon, P., H. El Housni, et al. (2006) The JAK2V617F Mutation Is Detectable at Very Low Level in Peripheral Blood of Healthy Donors. Leukemia 20(9): 1622.

Tefferi, A. and J.W. Vardiman (2008) Classification and Diagnosis of Myeloproliferative Neoplasms: The 2008 World Health Organization Criteria and Point-of-Care Diagnostic Algorithms. Leukemia 22(1): 14-22.