Cincinnati Children's

Division of Human Genetics

JAK2 V617F Quantitative Assay (JAK2)

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Additional information and test requisitions are available at:
www.cincinnatichildrens.org/molecular-genetics

Shipping Instructions

Please enclose an oncology test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:
Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229

The 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 a large percentage of cells from patients with chronic myeloproliferative disorders. V617F is an activating mutation.

This mutation can be helpful for clinicians in distinguishing between reactive cytosis and myeloproliferative disorders. Additionally, patients with this specific mutation might benefit from targeted therapies designed to inhibit the tyrosine kinase activity of JAK2.

INDICATION

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. Additionally, JAK2 V617F testing might aid in providing information that can be used to aid in treatment related decisions or in prognostication. These indications for testing are less well established than testing related to diagnosis.

TESTING 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.

ACCURACY:

The JAK2 MutaQuant™ assay is designed to detect less than 1 copy of JAK2 V617F mutation out of 100 normal copies of the gene, or a sensitivity of <1%. The variation of this assay though can be approximately 5% between laboratories and within a laboratory.

SPECIMEN:

PLEASE NOTE SAMPLE SHOULD BE RECEIVED SAME DAY OR SHIPPED OVERNIGHT ON ICE (DO NOT FREEZE)

5-10mls peripheral blood OR 3-5mls bone marrow in EDTA tube

TURN-AROUND TIME:

7-10 days

COST:

Please contact laboratory for pricing

CPT CODES:

83890, 83896, 83898, 83907, 83912, 83914

REFERENCES:

1. James C et al. (2005). Nature 434:1144.
2. Kralovics R et al. (2005). N Engl J Med 106:1779.
3. Lacout C, et al. (2006). Blood 108:1652.
4. Delhommeau F, et al. (2007). 109:71.