

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
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JAK2 Mutation Analysis

↑CPT

83890; 84311; 83898 (x2); 83896 (x2); 83912

↑Synonyms

Chronic Myeloproliferative Disorders (MPD); Janus Kinase 2 Gene (JAK2)

↑Test Includes

Relative quantitation of JAK2 V617F allele in human genomic DNA by real-time polymerase chain reaction.

↑Laboratory

Molecular Pathology

↑Availability

Monday-Friday 0700-1700

↑Turnaround Time

3-14 days

↑Specimen

Whole blood

↑Volume

3 mL

↑Minimum Volume

0.5 mL

↑ Container

Lavender top (EDTA) tube

↑ Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Specimens should be kept at room temperature or refrigerated but not frozen.

↑ Causes for Rejection

Clotted specimen; unlabeled or improperly labeled specimen; broken tube

↑ Reference Range

None

↑ Use

The V617F mutation in the Janus Kinase 2 gene (JAK2) was found to be present in several chronic myeloproliferative disorders (MPD), most frequently in polycythemia vera (65-97%), essential thrombocythemia (23-57%) and chronic idiopathic myelofibrosis (35-57%). This point mutation appears to cause constitutive activation of the JAK2 tyrosine kinase. Quantification of the JAK2 V617F mutation is useful both to assist in diagnosis of MPN, and for determining treatment and monitoring, as the aberrant tyrosine kinase appears to be a viable target for pharmacologic approaches.

↑ Methodology

The quantitative allele specific PCR technology is based on the use of specific forward primers for the wild type and the V617F allele, respectively. Only a perfect match between primer and target DNA allows extension and amplification in the PCR reaction. The increase in fluorescence signal is detected only if the target sequence is complementary to the probe and hence amplified during PCR. The number of PCR cycles necessary to detect a signal above the threshold is directly proportional to the amount of target present at the beginning of the reaction. Concentrations of V617F and wild type are determined by comparing the V617F and wild type crossing points to their standard curves. The relative concentration (%) of mutant cells in the specimen is calculated by comparing V617F target concentration to total target concentration.
