

# **JAK2 Mutation (V617F) Detection by Real -Time PCR**

## **Clinical Indication and Relevance**

- Confirms the diagnosis of myeloproliferative disorders (polycythaemia vera, essential thrombocythaemia, and primary myelofibrosis).
- Quantitation of *JAK2* V617F mutation load might be helpful in monitoring minimal residual disease.

## **Methodology**

Genomic DNA is isolated and amplified by allelic discrimination/quantitative real-time PCR targeting the *JAK2* gene. Results are reported as percentage of *JAK2* V617F mutant allele relative to the amount of wild type allele.

## **Sensitivity**

The assay sensitivity is 1% mutant DNA.

## **Turn-around Time**

Five to seven working days

## **Sample Requirements**

### **Collect**

- Peripheral blood (PB): 3-5 mL, in purple top (sodium EDTA) tube; yellow top (ACD) tube acceptable.
- Bone marrow (BM): 1-3 mL, drawn into a syringe containing anticoagulant and then delivered in purple top tube.

## **Transport**

Deliver immediately at 2-8°C (wet ice or cold packs). Do not freeze.

## **Stability**

Ambient - 1 hour; refrigerated - 48 hours.

## **Unacceptable Samples**

Serum or plasma; frozen PB or BM; clotted blood; severely hemolyzed samples.

## **CPT Code(s)**

81270: *JAK2* (Janus kinase 2) gene analysis, p.Val617Phe (V617F) variant

G0452-26: Molecular pathology procedure; physician interpretation and report

## **References**

1. Dupont S et al. Blood. 110:1013, 2007
2. Poodt J et al. Hematol Oncol. 24:227, 2006
3. Vannucchi AM et al. Leukemia. 21:1952, 2007
4. Wolstencroft EC et al. J Mol Diagn. 9:42, 2007