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

**Clinical Indication and Relevance**
- Confirms the diagnosis of myeloproliferative disorders (polycythemia vera, essential thrombocythemia, 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**