JAK2 Mutation (V617F) Detection by Real -Time PCR

Clinical Indication and Relevance

- Confirms the diagnosis of myeloproliferative disorders (polycythaemia vera, essential thombocythaemia, 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