Detection of JAK2 exon 12 Mutations

Clinical Indication and Relevance
Help in the diagnosis of myeloproliferative neoplasms, especially JAK2 V617F mutation negative polycythaemia vera.

Methodology
The assay is performed on patient genomic DNA by PCR amplification of JAK2 gene exon 12 region and Sanger sequencing. Mutation positive sample sequences are analyzed to determine mutation type. Results are reported as positive or negative for JAK2 exon 12 mutation.

Sensitivity
The assay’s sensitivity is 20% mutant DNA in a background of wild-type DNA.

Turn-around Time
Five to ten 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 (prefer 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)
81403 Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)

References