Detection of Somatic $MPL$ exon 10 Mutation

Clinical Indication and Relevance
Help in the diagnosis of myeloproliferative neoplasms, especially $JAK2$ and $CALR$ mutation negative essential thrombocythaemia and primary myelofibrosis.

Methodology
The assay is performed on patient genomic DNA by allele specific PCR amplification of $MPL$ gene exon 10 region, followed by fragment analysis. The presence and type of mutation is determined by mutant peak size. Results are reported as positive or negative for $MPL$ exon 10 mutation, and the type of $MPL$ mutation present.

Sensitivity
The assay’s sensitivity is 3% 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)
81402 Molecular pathology procedure, Level 3 (eg, >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis].

References