Somatic CALR exon 9 Mutation Detection

Clinical Indication and Relevance
- Help diagnosis of myeloproliferative neoplasms, especially JAK2 and MPL mutation negative essential thrombocythaemia and primary myelofibrosis.
- May be used as a prognostic marker for primary myelofibrosis, and thrombosis risk stratification for essential thrombocythaemia.

Methodology
Genomic DNA is isolated and amplified with specific primers targeting CALR exon 9 region. Amplified PCR products are then size fragmented on the ABI 3130x1 Genetic Analyzer. For CALR exon 9 mutation positive cases, DNA sequencing of the CALR exon 9 region is performed to characterize the mutation type. Results are reported as positive or negative for CALR exon 9 mutation, as well as mutation type.

Sensitivity
The fragment analysis assay’s sensitivity is 2% 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)
81479  unlisted molecular biology procedure

References