Myeloproliferative Disorders Panel (BCR-ABL, JAK2 V617F & Exon 12, MPL)

Test Code: 5040

Department: Molecular Oncology

CPT Code(s):
83890
83891
83894
83896 x 2
83898 x 6
83902
83903 x 2
83904 x 2
83909 x 2

Background:
Chronic myeloproliferative disorders (CMPDs) are clonal hematopoietic stem cell disorders characterized by proliferation of one or more myeloid cell lineages in the bone marrow and increased numbers of mature and immature cells in the peripheral blood. CMPDs include polycythemia vera (PV), essential thrombocythemia (ET), idiopathic myelofibrosis (IMF), and chronic myeloid leukemia (CML). These are broadly referred to as myeloproliferative disorders (MPDs).

While BCR-ABL translocation is the signature molecular derangement in chronic myeloid leukemia, most patients with other types of myeloproliferative disorders carry a V617F JAK2 mutation: 74 - 97% of patients with polycythemia vera (PV); 33 - 57% of patients with essential thrombocythemia (ET); 35 - 50% of patients with myelofibrosis (IMF); and also present infrequently (3-5%) in myelodysplastic syndrome (MDS) & CMML. In addition, molecular evidence of clonality for a subset of polycythemia vera cases lacking the V617F mutation includes a JAK2 exon 12 mutation. Similarly, the proportion of patients with ET or myelofibrosis lacking JAK2 mutation can be confirmed by a detection of mutation in the MPL gene.

To facilitate management planning for patients with chronic myeloproliferative disorders and to aid in the timely diagnosis of cases with equivocal clinicopathologic presentation, the myeloproliferative disorders panel offers analysis for four major molecular targets known to occur in specific chronic myeloproliferative disorder entities.

Clinical Utility of the Chronic Myeloproliferative Disorder Panel:
• To aid clinicopathologic confirmation of a specific CMPD entity
• To identify prognostically relevant clonal proliferation
• Targeted therapies for BCR-ABL are available, and may soon be available for JAK2

Methodology:
Separate platforms are used to detect different molecular targets in this panel. The detection of BCR-ABL is by quantitative real-time reverse-transcription polymerase chain reaction; the detections of JAK2 exon 12 mutations, and MPL mutation are by Sanger sequencing following target amplification; and the detection of JAK2 V617F is by allele-specific polymerase chain reaction.
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Sensitivity:
The BCR-ABL assay is capable of detecting target present at 0.01% abundance in a background of normal targets. The V617F JAK2 mutation is capable of detecting V617F at 0.1% abundance in a background of wild-type allele targets; and the JAK2 exon 12, and MPL mutation assays detect approximately 20% mutant allele in the background of wild-type allele.

Specificity:
A clinical correlation is imperative for the correct interpretation of this test panel.

Specimen Requirements:
- 5-10 mL of blood or bone marrow — yellow (ACD) or purple (EDTA) tube. Contact Client Services for shipping kit and instructions at (855) 535-1522. If sample cannot arrive at laboratory within 24 hours of draw, refrigerate until sample can be transported.

A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information.

Test Performed (Days):
Daily

Turnaround Time:
5-10 days

Shipment Sensitivity Requirements:
Keep specimen cold during transit, but do not ship on dry ice. Please use the cold pack provided in the KDL shipping kit. Ship the specimen overnight express, using the FedEx priority overnight label provided. The specimen must arrive at the lab no more than 24 hours after collection.

References:


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