POLYCTHEMIA VERA

London Health Sciences Centre | St. Joseph's Health Care London

Pathology and Laboratory Medicine

Orderable – E-order/Requisition

Turnaround Time: 1 month

Laboratory:
Molecular Diagnostics Lab

Requisition:
JAK2 REQUISITION / MOLECULAR DIAGNOSTICS REQUISITION

Method of Analysis:
JAK2 mutation, c.1849G>T, was detected using SNP allelotyping method by Agena MassARRAY (MALDI-TOF) platform. This technology uses single base extension reactions paired with mass spectrometry to quantitatively determine the identity of the amplified nucleotide based on the mass differential of the amplified nucleotides. Briefly, primers were designed using the MassArray Assay Design software. Amplified DNA was purified by SAP (Shrimp Alkaline Phosphatase). The primer was extended by one mass-modified nucleotide (ddNTP) specific for each

Alternate Name(s):

JAK2(V617F)

Specimen:

Whole blood-1 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

Interpretive Comments:

Myeloproliferative neoplasms (MPNs) are a group of diseases in which the bone marrow makes too many red blood cells, platelets, or certain white blood cells. Polycythaemia vera (PV), essential thrombocythaemia (ET), and primary myelofibrosis (PMF) are three main types of MPNs. The molecular pathogenesis of these disorders is unknown, but mutations of a tyrosine kinase gene, JAK2, have been implicated (PMID: 15781101, 15858187). It is known that a single point mutation in this gene, c.1849G>T, V617F, is present in 97% of patients with PV and 50-60% of patients with ET or PMF. Therefore, this specific mutation has been commonly used as a biomarker for molecular diagnosis, re-classification, and therapeutic intervention of these disorders (PMID: 25951317).

Storage and Shipment:

Must be received in testing laboratory within 5 days of collection, shipped at room temperature by courier/overnight delivery.

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assay designed. The cleaned products by resin were plated onto a 96-well SpectroCHIP® Array and then detected using the MassARRAY System (Agena). The software (MassARRAY typer 4) was configured to calculate the individual allele frequency for each SNP based on mass differential of each allele. Analytical sensitivity of this methodology is determined at 5% (by dilution of a JAK2 V617F-positive cell line into a negative cell line DNA). 5%-10% frequency is defined as indeterminate, with follow up testing of new specimen recommended; above 10% is defined as mutation detected.

Test Schedule:
As required,
Monday to Friday 0800-1600 hours

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