Patient Name:	**Dummy**	**CRM ID:**	NA
Age/DOB:	NA	**Sample Type:**	NA
Sex:	NA	**Collection date:**	NA
Referring Clinician:	N/A	**Received Date:**	NA
Test Requested:	**JAK2 Gene Analysis, 2 Exons (12 & 14) By Sanger Seq.**	**Reporting date:**	NA

JAK-2 (Exon14) Mutation Analysis by Sanger Seq.

CLINICAL DIAGNOSIS/SYMPTOMS

N/A

RESULTS

RESULTS	
JAK-2 (Exon 14)	**No variant Identified**

CLINICAL SIGNIFICANCE

JAK2 V617F mutations accounts for 90% PV patients and 60% of ET or MF patients. Rare Exon 12 insertion and deletion mutations in JAK2 accounts for 2-3% of PV. For the diagnosis of PV and ET three major criteria are defined of which one is the mutation in exon 12 or 14 (V617F) of JAK2 gene for PV and mutation in either JAK2, CALR or MPL gene for ET.

PV Patients with JAK2 exon 12 mutation have younger age, increased mean hemoglobin/hematocrit, and lower WBC & platelets count at diagnosis compared to those with JAK2 V617F mutation. However both JAK2 mutations are associated with similar rates of thrombo- sis, evolution to myelofibrosis or leukemia and death.

METHODOLOGY

Targeted sequencing and mutation analysis was performed by Polymerase Chain Reaction (PCR) followed by automated DNA sequencing of the amplicon using BigDye Terminator Chemistry on an ABI Genetic Analyzer 3500XL platform. Sequencing data were aligned to NCBI database to analyze the mutations

COMMENT

Chronic myeloproliferative neoplasms (MPNs) are clonal hematopoietic stem cell malignancies characterized by excessive production of blood cells. BCR-ABL1-negative MPN frequently harbor an acquired single nucleotide mutation in JAK2 characterized as c.G1849T; p. Val617Phe (V617F) and it is a gain-of-function mutation that leads to clonal proliferation. The JAK2 V617F is present in 95% to 98% of polycythemia vera (PV), and 50% to 60% of primary myelofibrosis (PMF) and essential thrombocythemia (ET). It has also been described infrequently in other myeloid neoplasms, including chronic myelomonocytic leukemia and myelodysplastic syndrome. Diagnostic criteria for ET, MF, and PV adopted by the World Health Organization (WHO) include identification of a clonal marker, with a specific recommendation to test for the JAK2 V617F mutation in exon 14. Detection of the JAK2 V617F is useful to help establish the diagnosis of MPN and The JAK2 allele burden decreases with successful therapy, disappears in some patients, and reappears during relapse.

LIMITATIONS

- A positive result is specific for a particular Myeloproliferative neoplasm (MPN) diagnosis and clinicopathologic correlation is necessary in all cases.
- A negative result does not exclude the presence of MPN or other neoplastic processes.
- The sensitivity of detection for Sanger sequencing is generally recognized as being approximately 15% to 20% mutant allele frequency.
- PCR is a highly sensitive technique; common reasons for paradoxical results are contamination during specimen collection, selec-tion of inappropriate specimen and inherent PCR inhibitors in the sample.

1. Tefferi A, Vardiman JW. Classification and diagnosis of myeloproliferative neoplasms: the 2008 World Health Organization criteria and point-of-care diagnostic algorithms. Leukemia. 2008;22:14-22.

2. Kiladjian JJ, Cassinat B, Turlure P, et al. High molecular response rate of polycythemia vera patients treated with pegylated interferon α-2a. Blood. 2006;108:2037-2040.

3. Kröger N, Badbaran A, Holler E, et al. Monitoring of the JAK2-V617F mutation by highly sensitive quantitative real-time PCR after allogeneic stem cell transplantation in patients with myelofibrosis. Blood. 2007;109:1316-1321.

4. Jerald Z. Gong, et al. Laboratory Practice Guidelines for Detecting and Reporting JAK2 and MPL Mutations in Myeloproliferative Neoplasms. J Mol Diagn 2013, 15: 733e744.

Conditions of Reporting

- Test results released pertain to the specimen submitted .
- All test results are dependent on the quality of the sample received by the Laboratory .
- Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician .
- Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours postreporting.
- Test results may show inter laboratory variations.
- If Sample collection date is not stated on test requisition form, the current date will be printed by default as the date of collection.
- Test results are not valid for medico legal purposes.

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.