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MC-6043

To: Narayana Superspeciality Hospital-Gurugram
Plot 3201, Block - V, DLF Phase - III,
Nathupur, Sector 24,
Gurugram,- 122002
Haryana
Report Of: LALA RAM

Sample ID	:	2460002524
Patient ID	:	1002431271
Collected on	:	05-06-2024
Received on	:	07-06-2024
Reported on	:	12-06-2024 05:58:16
Ref By	:	DR.RANDEEP SINGH

MOLECULAR GENETICS REPORT

Patient Name	: **LALA RAM**	**Age**	:	**72 Years**
Physician Name	: **DR.RANDEEP SINGH**	**Gender**	:	**Male**
Provisional Diagnosis	: **N.A.**	**Specimen Status**	:	**Ok**
Specimen Type	: **Bone Marrow Aspirate (BMA)**	**Disease Status**	:	**N.A**
Test Requested	: **c-KIT D816 and Exon 8 mutations, JAK2 V617F, JAK2 Exon12, CALR and MPL in Myeloproliferative Neoplasms**			

Test : JAK2 V617F, JAK2 Exon12, CALR and MPL in Myeloproliferative Neoplasms Analysis.

Result: Negative

JAK2 V617F, JAK2 Exon12, CALR and MPL in Myeloproliferative Neoplasms Analysis:

Gene	Exon	Codon	Result	Mutation	COSMIC ID	Clinical significance
JAK2	14	V617	Negative	-	-	-
JAK2	12	-	Negative	-	-	-
MPL	10	W515	Negative	-	-	-
CALR	9	-	Negative	-	-	-

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LALA RAM Sample ID: 2460002524

Conclusion: The specimen tested negative for JAK2 V617F mutation, JAK2 exon 12 mutations, CALR exon 9 mutations and MPL W515K/L mutation. Kindly correlate with clinical findings.

Interpretation:
Myeloproliferative neoplasms (MPN) frequently harbor the JAK2 V617F mutation in exon 14 in more than 95% of patients with Polycythemia Vera (PV), and about 60% of patients with essential thrombocytosis (ET) and primary myelofibrosis (PMF) Literature reports indicate a lower incidence of exon 12 mutations in the western population as compared to the Asian population suggesting an uneven geographic distribution. The CALR mutation frequencies in patients with ET and PMF were around 19% and 22%, respectively. The MPL mutation frequencies in patients with ET and PMF were around 6% and 10%, respectively.

Methodology:
DNA was extracted from the specimen and JAK2 V617F mutation analysis was carried out using allele-specific polymerase chain reaction which can detect both wild type and mutant allele in the presence of an internal control which is a fragment of the JAK2 sequence. The analytical sensitivity of the assay has been determined to be 5% of tumor load. DNA reference sequence is NM_001322194.1, protein reference sequence is NP_001309123.1. The MPL mutant is detected by allele-specific polymerase chain reaction which can detect the wild-type allele as well as the two mutant alleles i.e. W515L and W515K; JAK2 Exon 12 mutation and CALR mutation were detected by DNA sequencing using capillary electrophoresis of their respective coding regions. The analytical sensitivity is determined to be 15% of the tumor load. Reference sequences for MPL: NM_005373.2 & NP_005364.1; for CALR: NM_004343.3 & NP_004334.1

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References:

1. Labastida-Mercado. et al. The mutation profile of JAK2, MPL and CALR in Mexican patients with philad elphia chromosome-negative myeloproliferative neoplasms. Hematol Oncol Stem Cell Ther 2015; 8(1) : 16-21.
2. Teofili et al. Markers of Myeloproliferative Diseases in Childhood Polycythemia Vera and Essential Thr ombocythemia. J Clin Oncol.2007; 25:1048-1053.
3. Pietra D. et al. Differential clinical effects of different mutation subtypes in CALR-mutant myeloprolife rative neoplasms. Leukemia 2016;30(2):431-438.
4. Rumi E, et al. Clinical effect of driver mutations of JAK2, CALR, or MPL in primary myelofibrosis. Blood. 2014;124(7):1062-9.

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- End of Report -

Conditions of Reporting/Disclaimer:

- This report is based on the sample received in the Lilac Insights laboratory; the analysis is based on the assumption that samples received are representative of the patient mentioned on the test requisition form and the sample. When samples are received from various referral centers, it is presumed that patient demographics are verified at the point of sample collection.
- All samples for molecular studies must be collected in EDTA tubes (lavender cap). A sample where the test requisition requires RNA and subsequent cDNA conversion, must be maintained and transported at 4°C until it reaches Lilac Insights Pvt. Ltd. within 24 hours to prevent degradation of the RNA in the sample. FFPE tissue blocks often yield fragmented DNA of low concentrations which can impact result quality.
- Assays are performed in accordance with standard procedure on receipt. The reported results are dependent on individual assay methods, equipment used, method specificity, sensitivity and quality of specimen(s) received.
- Despite all the necessary precautions and stringency adopted whilst performing DNA tests, the currently available data indicates that the technical error rate associated with all types of DNA analysis, is approximately 2%.
- Although molecular testing is highly accurate, rarely false-positive & false-negative diagnostic errors may occur due to improper quality control during sample collection, cellular integrity of sample, selection of inappropriate specimen and/or presence of PCR inhibitors. PCR primer binding site polymorphisms or mutations might lead to allele dropout & cause false negative results.
- It is important that all clinicians or persons requesting DNA diagnostic tests are aware of these data before acting upon these results. As with all diagnostic tests, the laboratory report must be interpreted in conjunction with the presenting clinical profile of the patient and evaluation of all reports.
- In sequencing based tests sometimes variants of unknown significance (VUCS) are detected that have either not been reported before, and/or whose effect cannot be determined based on the current knowledge standards and reporting guidelines. In such cases, we recommend periodic review of these variants to determine any change in classification based on new published research.
- Lilac Insights Pvt. Ltd. has policy to return the FFPE blocks within one month after final reporting with proper documentation of the dispatch of the block to customer from accession dept, Lilac. After dispatch, if there is no intimation from customer within two weeks, Lilac will not be responsible for the dispatched FFPE block.
- Soft copies of reports are sent to customer by office mail ID. Also, Hardcopies are sent to customer only on the address provided by client.

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