ISO 9001 : 2008 CERTIFIED LAB

KOS Diagnostic Lab
(A Unit of KOS Healthcare)

KOS™
EXCELLENCE IN HEALTHCARE & DIAGNOSTICS

Dr. Vinay Chopra
MD (Pathology & Microbiology)
Chairman & Consultant Pathologist

Dr. Yugam Chopra
MD (Pathology)
CEO & Consultant Pathologist

Patient Details

Name	RAJ PAL	**Sex / Age**	M / 31 Years	**Case ID**	30521601075
Ref By		**Dis.Loc.**		**PT. ID**	
Test Name	JAK2 Mutation Panel (Exons 12 to 15)	**Bill. Loc.**	KOS DIAGNOSTIC LAB	**PT. Loc.**	
Disease of Interest (DOI)	Myeloproliferative neoplasm(MPN)				

Sample Details

Registration Date & Time	2023-05-16 15:56:25	**Sample Type**	Whole Blood EDTA	**Ph # Re**	-
Sample Date & Time	2023-05-16 15:56:00	**Ref ID 1.**	-	**Acc. Remarks**	-
Report Date & Time	2023-05-30 09:57:09 AM	**Ref ID 2**	-	**PT. Loc.**	KOS DIAGNOSTIC LAB

Clinical History

Suspected case of Myeloproliferative neoplasm(MPN)

Test Results and Interpretation

NO CLINICALLY SIGNIFICANT MUTATION DETECTED IN JAK2 GENE

Recommendations

- **The patient is negative for mutations in JAK2 gene (exons 12-15 including V617F).**
- **Please note: Analysis was focused on the gene requested.**
- **Please correlate with clinical features, CBC, bone marrow findings and cytogenetics for final conclusion.**

Technical Notes

Methodology: Massively Parallel Sequencing (Next Generation Sequencing) Genomic DNA from the submitted specimen was enriched for the complete coding regions and splice site junctions of genes listed below using a custom bait-capture system. Paired-End Sequencing was performed with 2x100/2x150 chemistry, on an Illumina platform. Reads were assembled and were aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. Data was filtered and analysed to identify variants of interest and interpreted in the context of a single most damaging, clinically relevant transcript for the purpose of the report, indicated as a part of variant details. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 5-10bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design.

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Patient Name : RAJ

Approved by Dr. Arpan Mehta
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NOTE:
This Sample was outsourced

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Deletion and duplication analysis is performed in cases when indicated but detected variations need to be confirmed by an alternate methodology. Variants are reported according to the Human Genome Variation Society (HGVS).

Panel: JAK2 Mutation Panel (Exons 12 to 15): The assay is focused only on the variants found in the JAK2 gene(Exons 12 to 15).

Tools and Databases employed for analysis: Clinvar, UCSC genome browser, gnomAD, Pubmed, VARSOME, various bioinformatics analysis, predictive tools, and disease-specific databases used as available and appropriate.

Hotspot Genes		Full Genes
24 genes		16 genes
DNA		
ABL1	KRAS	ASXL1
BRAF	MPL	BCOR
CBL	MYD88	CALR
CSF3R	NPM1	ETV6
DNMT3A	NRAS	EZH2
FLT3	PTPN11	IKZF1
GATA2	SETBP1	NF1
HRAS	SF3B1	PHF6
IDH1	SRSF2	PRPF8
IDH2	U2AF1	RB1
JAK2	WT1	RUNX1
KIT	CEBPA	SH2B3
		STAG2
		TET2
		TP53
		ZRSR2

Test Limitations

- Predisposition to disease may be affected by non-genetic factors.
- Variants of hitherto unknown significance may be detected and may not be reported subject to analysis by various methodologies.
- Although molecular testing is highly accurate, rarely false-positive & false-negative diagnostic errors may occur.
- Only mutations having VAF >5% (except JAK2 V617F) will be reported.
- PCR primer binding site polymorphisms or mutations might lead to allele dropout & cause false negative results.
- This test is not designed for the detection of minimal residual disease.
- Mutations in underperforming amplicons (<100x coverage) may be missed.
- Indels exceeding 50 bp size may not be detected.
- All laboratory tests are associated with an error rate of ~1%. These could be due to sample mismatch, inappropriate labeling, processing, or technological limitations.
- Please correlate with clinical features and other investigations for the final conclusion and send a repeat sample for analysis if necessary.
- This report should not be copied or reproduced except in its entirety.
- For tests performed on specimens received or collected from non-NCGM locations, it is presumed that the specimen belongs to the patient named or identified as labeled on the container/test request and such verification has been carried out at the point of generation of the said specimen by the sender.
- These test results should be interpreted by a Physician only in conjunction with the patient's clinical history, other test results and any previous analysis of appropriate family members.
- Variants of unknown significance are mutations detected on sequence analysis that have either not been reported before, or whose effect cannot be determined based on the current knowledge standards and reporting guidelines. We recommend periodic review of variants of unknown significance on an annual basis to determine any change in classification based on new published research.
- Unless reported or predicted to cause disease, alterations found that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

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- The classification and interpretation of all the variants in this assay reflects the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific information comes to light. We recommend re-analysis of this report yearly, in order to take advantage of any new scientific data that may become available. A fee may apply for this service.

Reviewed By

Dr. Arpan Mehta
MD Pathology
PDF Lab Haematology(TMC Kolkata:1yr)
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