MEDGENOME

Sy No 94/1C and 94/2, Tower-1, Veerasandra Village, Attibele Hobli, Electronic City, Phase-1, Bangalore, Karnataka, 560100.

Fluorescent In Situ Hybridization (FISH) REPORT: FISH for Myeloproliferative Neoplasms (MPN) panel - 4 markers (PDGFRA, PDGFRB, JAK2 & FGFR1 Gene Rearrangements)

Patient Name	H.L.M Lakvindu	**Requesting Clinician**	Dr. Mahendra Perera
Gender	Male	**Hospital Information**	Aegle Omics Private Limited
Age/Date of Birth	24 Years	**Sample Source**	Peripheral Blood in Sodium Heparin.
Sample ID	9361512	**Samples Collected(Date & Time)**	27-08-2025 3:54 pm
Order ID(s)	1428771	**Samples Received(Date & Time)**	30-08-2025 6:00 pm
Clinical Indication	Case of MPN.	**Report Date**	05-09-2025 1:20 pm
Collection Center/ Partner Lab	0		

RESULT SUMMARY

Sl no	Probe Name	FISH Result	ISCN 2024
1	PDGFRA GENE Deletion/rearrangement	Negative	nuc ish(PDGFRA)x2[198/200]
2	JAK2 gene rearrangement	Negative	nuc ish(JAK2)x2[198/200]
3	PDGFRB Gene rearrangement	Negative	nuc ish(PDGFRB)x2[198/200]
4	FGFR1 gene rearrangement	Negative	nuc ish(FGFR1)x2[198/200]

Interpretation:

FISH test is negative for PDGFRA Deletion/rearrangement, PDGFRB rearrangement, FGFR1 rearrangement and JAK2 rearrangement. Kindly correlate with clinical presentation.

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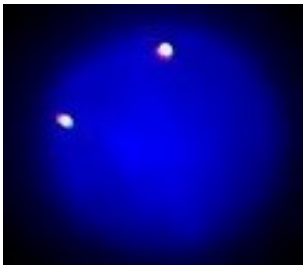
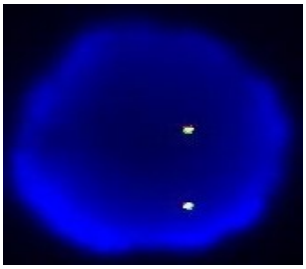
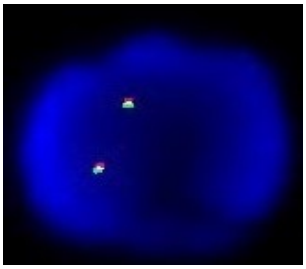
DETAILED REPORT



PDGFRB Gene rearrangement



JAK2 gene rearrangement



FGFR1 gene rearrangement

1) PDGFRB :

Spectrum Green (G)	**Positioned telomeric to the PDGFRB gene.**	**Spectrum Orange (O)**	**Positioned centromeric to the PDGFRB gene.**

Loci Analyzed	Signal Pattern	Normal Cut Off (%)	Percentage of Cells showing signal pattern	Result
PDGFRB	**2F/0G/0O**	**>= 98 %**	**99 %**	**Negative**

2) JAK2 :

Spectrum Green (G)	**5' of JAK2(9p24)**	**Spectrum Orange (O)**	**3' of JAK2(9p24)**

Loci Analyzed	Signal Pattern	Normal Cut Off (%)	Percentage of Cells showing signal pattern	Result
JAK2	**2F/0G/0O**	**>= 98 %**	**99 %**	**Negative**

3) FGFR1 :

Spectrum Green (G)	**5'of FGFR1 (8p11)**	**Spectrum Orange (O)**	**3' of FGFR1 (8p11).**

Loci Analyzed	Signal Pattern	Normal Cut Off (%)	Percentage of Cells showing signal pattern	Result
FGFR1	**2F/0G/0O**	**>= 98 %**	**99 %**	**Negative**

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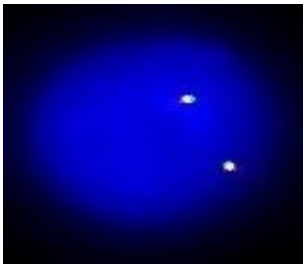

PDGFRA GENE Deletion/rearrangement

4) PDGFRA :

Spectrum Green (G)	**Proximal to FIP1L1,PDGFRA and the region distal to the gene**	**Spectrum Orange (O)**	**hybridizing CHIC2 gene region**

Loci Analyzed	Signal Pattern	Normal Cut Off (%)	Percentage of Cells showing signal pattern	Result
PDGFRA	**2F/0G/0O**	**>= 98 %**	**99 %**	**Negative**

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Testing methodology: FISH is a molecular cytogenetic technique used to detect the presence or absence and location of specific gene sequences. FISH involves co-denaturation and hybridization of fluorescent labelled specific DNA probes to target DNA sequence in the interphase cells. The excess unbound probe is removed during post hybridization washes. The sample is stained with a DAPI (4',6-Diamidino-2-phenylindole) counter-stain to demarcate the nuclei. Each fluorescent labelled probe that hybridizes to region of interest in interphase cells is visualized as signal using suitable optical filters under Epi fluorescent microscope. 200 interphase cells were counted for each probe manually by two readers. Interpretation of results are done based on the signal patterns observed. Based on these interpretations, the results of the test are reported. Metasystems probes used for this panel. Appropriate controls are run in each batch along with patient samples.

The updated (2017) World Health Organization (WHO) classification of tumors of hematopoietic and lymphoid tissues indicates the category myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB, FGFR1. The most common rearrangement of PDGFRB is ETV6-PDGFRB, but more than 20 fusion partners have been identified. Myeloid neoplasms (MPNs) with rearrangement of PDGFRB are phenotypically and genotypically diverse. MPNs associated with rearrangement of PDGFRB are responsive to Imatinib.

References:

1. WHO Classification of tumors of hematopoietic and lymphoid tissues, Revised edition 2017.
2. WUhan healthcare probe kit insert.

Disclaimers:

1. This test was developed, and its performance characteristics determined by MedGenome. It has not been cleared or approved by the US Food and Drug Administration.
2. The finding of this test must be correlated with other clinical, haematopathological and cytogenetic findings for complete analysis.
3. Genetic changes other than those assayed cannot be ruled out on the basis of this testing.

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End of Report