Dean Diagnostics
DIAN DIAGNOSTICS

Dean Medical Laboratory Test Report
DIAN Medical Laboratory Test Report
Molecular Pathology Test Report

Submitting Institution: Shanghai Songjiang District Central Hospital **Barcode No.: 990129374506**

Name: Miao Yindi	Patient Category: Inpatient	Department: Hepatology Ward	Test ID: B2025-3470
Gender: Female	Patient Phone:	Bed No.: 5306	Sample Type: Bone Marrow
Age: 79 years	Referring Physician: Zhu Ruifeng	Outpatient/Inpatient ID: 75000630251	Sample Status: Normal appearance

Clinical Diagnosis: Infectious fever, Gallstones, Hypertension Grade 1 (high-risk), Thrombocytopenia, Kidney stones

Primary Testing Equipment: ThermoBrite、NIKON、Axio Imager.Z2/Metafer

Testing Method: fluorescence in situ hybridization

Sample ID:B2025-3470

Paraffin Block No.:/

Test Results

Detection probe	Reagent Manufacturer	Red Locus (R)	Green Locus (G)	Abnormal Signals and Ratios	Threshold	Conclusion
PDGFRA Break	Ambiping® (retained as proper noun/brand name)	CHIC2(4q12)	FIP1L1, PDGFRA(4q12)	/	0.57%	Negative
PDGFRB Break	Ambiping® (retained as proper noun/brand name)	3'PDGFRB(5q32)	5'PDGFRB(5q32)	/	0.71%	Negative

*: See remarks for details

Result Description:

nuc ish(PDGFRA,CHIC2,FIPL1x2)[500]
nuc ish(PDGFRBx2)[500]

Test Conclusion:

All the above loci were negative.

Laboratory Technologist: 武家瑶 Reviewer: Approver: Same

Testing Laboratory: Hangzhou Dian

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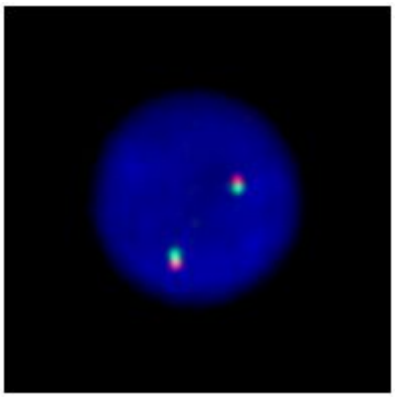
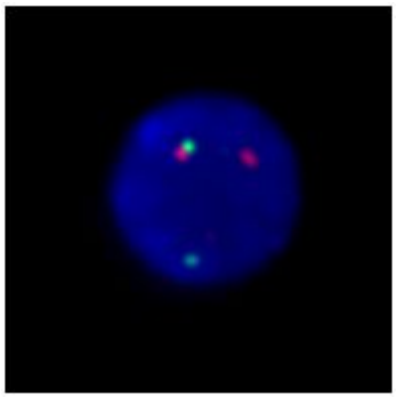
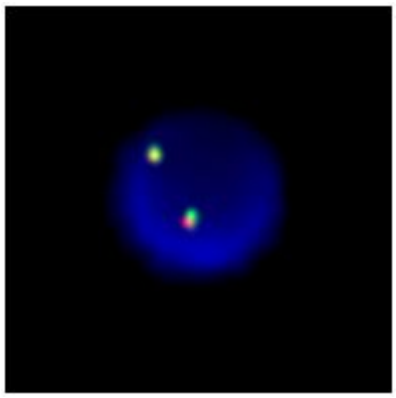
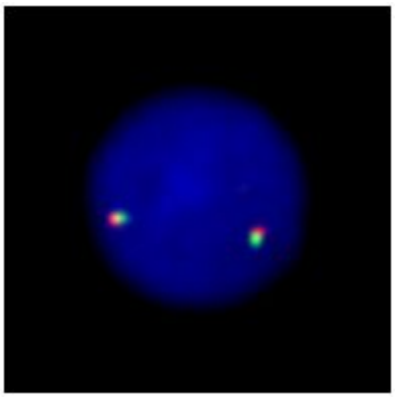
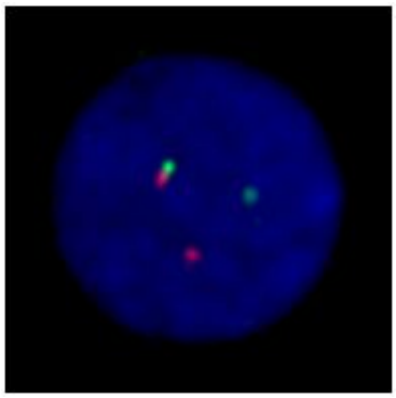
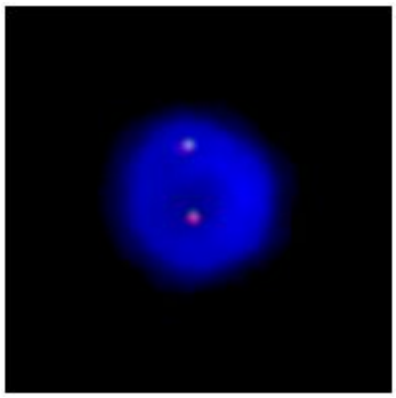
Test diagram

Detection probe	Negative control	Positive control	Test image
PDGFRA Break			
PDGFRB Break			

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Interpretation reference

Detection probe	Clinical significance	References
PDGFRA break	The PDGFRA (platelet-derived growth factor receptor, alpha polypeptide) gene is located at 4q12. Its encoded protein, platelet-derived growth factor receptor alpha, is a cell surface receptor tyrosine kinase. Upon binding with its corresponding ligands such as PDGF, it becomes activated and subsequently triggers the phosphorylation pathways of phosphatidylinositol, cAMP, and various proteins, regulating cell division and proliferation. Abnormal gene activation can lead to tumorigenesis and promote tumor angiogenesis. In hematologic diseases, various fusion genes/chromosomal rearrangements that cause abnormal activation of PDGFRA have been identified. The most common is the FIP1L1-PDGFRA fusion, which results from a cryptic deletion involving the CHIC2 gene region between FIP1L1 and PDGFRA on chromosome 4 (cryptic del(4)(q12)). The WHO Classification criteria (2016) categorize 'Myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB, or FGFR1, or with PCM1-JAK2' as a distinct major class, and further list 'Myeloid/lymphoid neoplasms with PDGFRA rearrangement' as one of its subclasses. Myeloid or lymphoid neoplasms with PDGFRA rearrangement respond well to imatinib , but cases of imatinib resistance due to PDGFRA gene mutations (commonly T674I) have been reported.	[1] Pardanani, A., et al. Blood, 2003. 102(9): p. 3093-6. [2] Pardanani, A., et al. Leuk Res, 2006. 30(8): p. 965-70. [3] Tefferi, A. Acta Haematol, 2005. 114(1): p.52-60. [4] Leukemia & Lymphoma Group, Chinese Society of Hematology. Chinese Journal of Hematology, 2017, 38(7): p. 561-565. [5] Arber, D.A., et al. Blood, 2016. 127(20): p. 2391-405.
PDGFRB break	The PDGFRB (platelet-derived growth factor receptor, beta polypeptide) gene is located at 5q32. Its encoded protein, platelet-derived growth factor receptor β, is a cell surface receptor tyrosine kinase that activates upon binding with ligands such as PDGF, regulating cell division and proliferation. Abnormal gene activation can lead to tumorigenesis and a series of clinical abnormalities. In hematologic diseases, PDGFRB rearrangement is typically caused by the t(5;12)(q32;p12) translocation resulting in ETV6-PDGFRB fusion, but not all t(5;12)(q32;p12) carry the ETV6-PDGFRB fusion gene. In addition to the most common ETV6-PDGFRB , there are at least 20 other partner genes that can form fusion genes with PDGFRB. The WHO classification criteria (2016) classified "Myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB, or FGFR1, or with PCM1-JAK2" as a distinct major category, and listed "Myeloid/lymphoid neoplasms with PDGFRB rearrangement" as one of its subcategories. Myeloid or lymphoid neoplasms with PDGFRB rearrangement respond well to imatinib treatment.	[1] Tefferi, A. Acta Haematol, 2005. 114(1): p.52-60. [2] Leukemia & Lymphoma Group, Chinese Society of Hematology. Chinese Journal of Hematology 2017, 38(7): p.561-565. [3] Passamonti, F. Blood, 2014. 123(23): p.3526-8. [4] Cheah, C.Y., et al. Blood, 2014. 123(23): p. 3574-7.

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Testing Declaration

1. This test result is valid only for the submitted sample.

2. This test result only reports genetic variants within the detection scope. A negative result does not exclude the presence of other genetic mutations outside the detection range.

3. This test is primarily intended to assist clinical decision-making. Medication recommendations are for physician reference only and do not constitute medical advice. Specific treatment plans must be determined by clinicians.

4. This test result is time-sensitive. Since tumor progression is a dynamic process, strictly speaking, this test only reflects the gene mutation status of the patient's tumor at the time of sample collection.

5. Our center reserves the final right of interpretation for the above test results. If you have any questions, please contact us within 7 working days after receiving the results.

Laboratory Technologist: 武家瑶 Reviewer: Approver: Same

Testing Laboratory: Hangzhou Dian

检验专用章

Report Service

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