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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: KAILASH [157658]

Sample ID	:	2460002509
Patient ID	:	160245440
Collected on	:	07-06-2024
Received on	:	08-06-2024 18:40:00
Reported on	:	12-06-2024 05:53:39
Ref By	:	Narayana Superspeciality Hospital-Gurugram

MOLECULAR CYTOGENETICS (FISH) REPORT

Patient Name	: **KAILASH [157658]**	**Age**	: **82 Years**
Physician Name	: **DR. RANDEEP SINGH**	**Gender**	: **Female**
Provisional Diagnosis	: **?Myeloma**	**Specimen Status**	: **Ok**
Specimen Type	: **Bone Marrow Aspirate (BMA)**	**Disease Status**	: **N.A**
Test Requested	: **Multiple Myeloma on purified plasma cells FISH, Conventional Karyotype**		

Test	: **Multiple Myeloma on purified plasma cells OncoInsights™ FISH panel**
Test panel	: Monosomy13/del(13q), *TP53* (17p) deletion, 1q amplification and 1p deletion, Trisomy 3, 7, 9 & 15, *IGH* (14q32) translocation Analysis.
Method	: Isolation and purification of CD138 sorted plasma cells from bone marrow mononuclear cells, followed by Fluorescence in situ hybridization and analysis on plasma cells.
Probe panel	: ZytoLight SPEC *CKS1B/CDKN2C* Dual Color, SPEC *RB1*/13q12 Dual Color Probe, SPEC *TP53* (17p13) / CEN 17 dual color, CEN 3, CEN 7, SPEC *IGH* Break Apart, SPEC *IGH/MYC* DF, Metasystem XL 5p15 / 9q22 / 15q22 Hyperdiploidy probe. Limit of Detection: Dual fusion probe: ≤1% , Break apart probe: 5%, Centromeric probe: 2% (trisomy), 5% (monosomy), LSI deletion probe: 5%, LSI Amp (1q): 3-5%
No. of Cells Analysed	: 200

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Result:
***IGH* (14q32) translocation Analysis:**

***IGH*::? (14q32) Translocation**	***IGH* (Orange/Green)**	**5` *IGH* (Green)**	**3` *IGH* (Orange)**	**No. Cells**
Signal/s/Cell	2	0	0	200

Monosomy13/del(13q),*TP53* (17p) deletion,1q amplification and 1p deletion Analysis:

-13/del(13q)	**RB1 (13q14.2) (Orange)**	**13q12.11 (Green)**	**No. Cells**
Signal/s/Cell	2	2	200
17p deletion	***TP53* (Orange)**	**CEN 17(Green)**	**No. Cells**
Signal/s/Cell	3	3	16/200
1q amp/1p del	***CDKN2C*(1p32.2) (Green)**	***CKS1B*(1q21) (Orange)**	**No. Cells**
Signal/s/Cell	2	3	14/200

Trisomy 3, 7, 9 & 15 Analysis:

	Signal/s/Cells	**No. Cells**
CEN 3 (Orange)	3	10/200
XL 5p15 (Green)	4	14/200
CEN 7(Orange)	3	12/200
XL 9q22 (Aqua)	3	14/200
XL 15q22.3 (Orange)	3	14/200

Interpretation: Fluorescence in situ hybridization (FISH) showed evidence of 1q gain (Freq. 7%).
Signal pattern showed evidence of trisomy of chromosome 3 (Freq. 5%), tetrasomy of chromosome 5 (Freq. 7%), trisomy of chromosome 7 (Freq. 6%), trisomy of chromosome 9 (Freq. 7%), trisomy of chromosome 15 (Freq. 7%) and trisomy of chromosome 17 (Freq. 8%). Gain of chromosome 3, 5, 7, 9, 15 and 17 revealed hyperdiploidy.
There was no evidence of *IGH*::? translocation, monosomy 13/del(13q), 17p deletion and 1p deletion. **Additional FISH study with *IGH*/*MYC* probe showed no evidence of *IGH::MYC* translocation.**
Impression: Present case revealed hyperdiploidy with 1q gain. 1q gain is intermediate-high risk abnormality whereas hyperdiploidy is low risk abnormality in MM as per reported lit.

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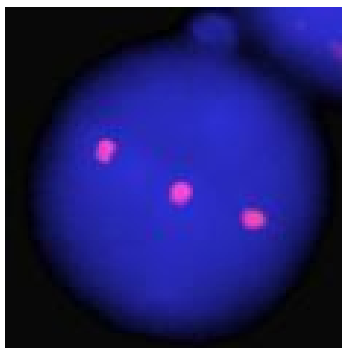
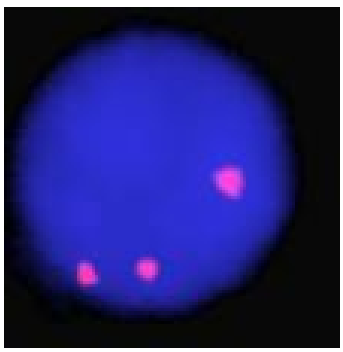


FISH on interphase cells showing trisomy of chromosome 3



FISH on interphase cells showing trisomy of chromosome 7

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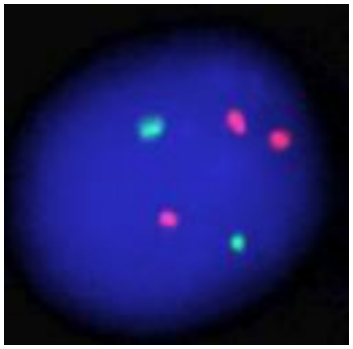

FISH on interphase cells showing 1q gain

Prepared By : **Mahima Patil**
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- End of Report -

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- The report relates only to the specimen submitted to the lab which was verified and confirmed at the time of specimen collection. Also it is presumed that the specimen belongs to the patient named or identified, such verification being carried out at the point of generation of the said specimen.
- Although Conventional karyotyping is a gold standard method of cytogenetics which gives a global whole genomic view of multiple known, unknown chromosomal abnormalities, small cryptic, subtle aberrations below 7-8 Mb resolution can be missed.
- In spite of known sensitivity and efficiency of the genetic test, the test results have to be correlated with other clinical and pathological finding for conclusive diagnosis and disease management.
- A test request may be revised or generated by Lilac geneticist with an intimation to an Oncologist if: 1) Incomplete requisition 2) After haematopathology Update.
- In 1-2 % of APL cases, FISH may turn out to be negative due to PML/RARA probe design which unable to detect cryptic insertion of PML to RARA. In such rare cases, It is advisable to check PML-RARA by molecular methods.
- In case of Multiple Myeloma, flowcytometry report indicating abnormal plasma cell population is important for reference, as small abnormal clones may get deduced as per limit of detection policy in FISH analysis.
- In case of FFPE FISH, if H & E stained slides &/or histopathology report is not provided by customer , LILAC proceed with H & E staining followed by histopathology remarks along with marking of tumor area by our consultant pathologist.
- 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.
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