DEPARTMENT OF CYTOGENETICS

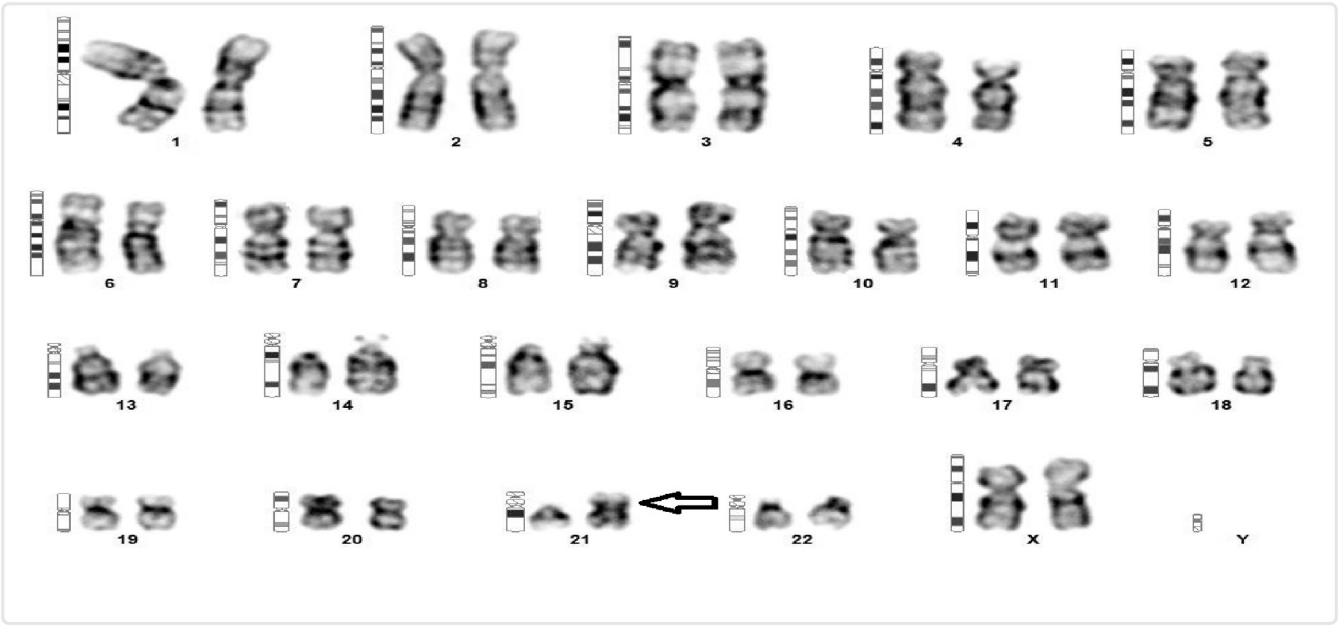
Patient Name	**B/O. KAVYA**	**Reg.No**	**0352407070019**	**Barcode**	**A0577358**
Age/Gender	**26 D / FEMALE**	**-**	**-**	**Collected Date**	**07-07-2024 02:50 PM**

CHROMOSOMAL ANALYSIS-KARYOTYPING

SAMPLE RESULTS	
Sample Type	Peripheral Venous Blood.
Quality of Sample	Good.
Yoda Cytogenetics Number	CYG-24-PB-1204
Clinical Indication	Not provided
Test Requested	Karyotyping on Blood sample.
Test Methodology	Stimulated Peripheral Blood lymphocyte culture.
No of cells counted	20
No of cells Karyotyped	05
Estimated band resolution	400-500 bphs
Banding method	GTG
ISCN	2020

Initial

CYTOGENETICS REPORT -CYG-24-PB-1204



DEPARTMENT OF CYTOGENETICS

Karyotype

46,XX,der(21;21)(q10;q10),+21

Interpretation

Chromosomal analysis of PHA stimulated Peripheral Blood lymphocytes revealed a female chromosomal complement with the presence of robertsonian translocation between the long arms of homologous chromosomes 21 along with the presence of an additional copy of chromosome 21 in all cells analyzed. This karyotype is suggestive of a female child with Translocation Down Syndrome. Kindly correlate cytogenetic findings with clinical features.

Recommendations

1.Parental karyotyping is suggested to rule out or confirm familial inheritance of translocation.
2.Genetic counseling is recommended for patient management.

References

1. An International System for Human Cytogenetic Nomenclature (2020). Karger Publishers.
2. Human Cytogenetics: Constitutional Analysis. A Practical Approach. Third Edition, Edited by Denise Rooney.

Disclaimer

1. This assay allows for microscopic visualization of numerical and structural abnormalities.
   The limitation in size of the chromosomal abnormalities like deletion(interstitial and terminal) , translocation, inversion , duplication(interstitial and terminal) and other structural aberrations size from >5mb to 10mb.
2. Sample received without relevant clinical history , family history , previous medical reports related to cytogenetics and microarray will not be accepted .The lab is not responsible for any deviation in interpretation of the assay as a result of not being provided the necessary relevant clinical information.
3. Sample not received in appropriate containers or not collected optimally may lead to poor GTG banding and low resolution and therefore have high chances of missing structural abnormalities. The lab does not address the problems related to inappropriate sample collection and handling (preanalytic issues)
4. Test results are based on the sample received in the department and the results and interpretation are in the context of the demographic details received along with the sample.
5. Reporting TAT may be delayed due to the unsought circumstances and extra workout and repeat culture and clinical correlation with other parameters.
6. Test results are reported as per updated and current version of ISCN.
7. Partial reproduction of the report is not permitted
8. The content of the report may be used for research purpose without revealing the personal information of the subject.
9. Detection of heterogeneity of the clonal cell population in the specimen (i.e., mosaicism) is limited by the number of cells analyzed and karyotypes per report.