Karyotyping by G-Banding Peripheral Blood

INTERNAL LAB NO.	962-22-K
CULTURE METHOD	72-hour stimulated cultures were put up with appropriate mitotic agents.
BANDING METHOD(S)	GTG-Banding with Trypsin & Giemsa with 450-550 bands pattern (ISCN-2016).
CLINICAL INDICATION(S)	? Down Syndrome,
NO.OF CELLS COUNTED	20
NO.OF CELLS ANALYZED	20
NO.OF CELLS KARYOTYPED	10
KARYOTYPE RESULT	**46,XY,der(14;21)(q10;q10),+21**
INTERPRETATION	The Karyotype contains one normal chromosome 14, two normal chromosome 21, and der (14; 21).
COMMENTS	Report consistent with Down syndrome (Robertsonian Translocation).The resulting net imbalance is loss of the short arm of chromosome 14 and trisomy for the long arm of chromosome 21 indicated by 46 count.
RECOMMENDATION(S)	Kindly correlate clinically. In case of any discrepancy kindly send new sample for confirmation. Along with Consultation with pediatrician for growth and management of the child Genetic Counseling (Test code G0078) and prenatal testing is recommended for future pregnancy. For any queries please feel free to contact at Department of Medical Genetics on 022-43560767.

Karyotype analysis detects all numerical and gross structural anomalies within the limits of the assay procedure. Microdeletions,microduplications, single gene disorders and low grade mosaicismhowever would not be ruled out. FISH/CMA/Molecular studies are recommended for the same.Clinical correlation is advised.

Note: Importance of Clinical Indicatons

1. Clinical details/history findings including age and sex of patient are important for accurate selection of culture method
2. Clinical details to be provided in the form of ultrasound information / phenotypic features / family history, etc.
3. For investigation of mosaicism which requires screening of large number of metaphase cells.
4. To target analysis for a particular chromosome in the form of high resolution banding.
5. For recommendation of further investigation - eg: FISH, Molecular Genetics Studies. Genetics abnormalities like single gene / polygenic disorders, microdeletions, subtle rearrangements, low grade mosaicism may not be detected by G-Band Karyotyping and may require more sensitive testing like FISH and Chromosomal Microarray.

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NOTE:
This Sample was Outsourced

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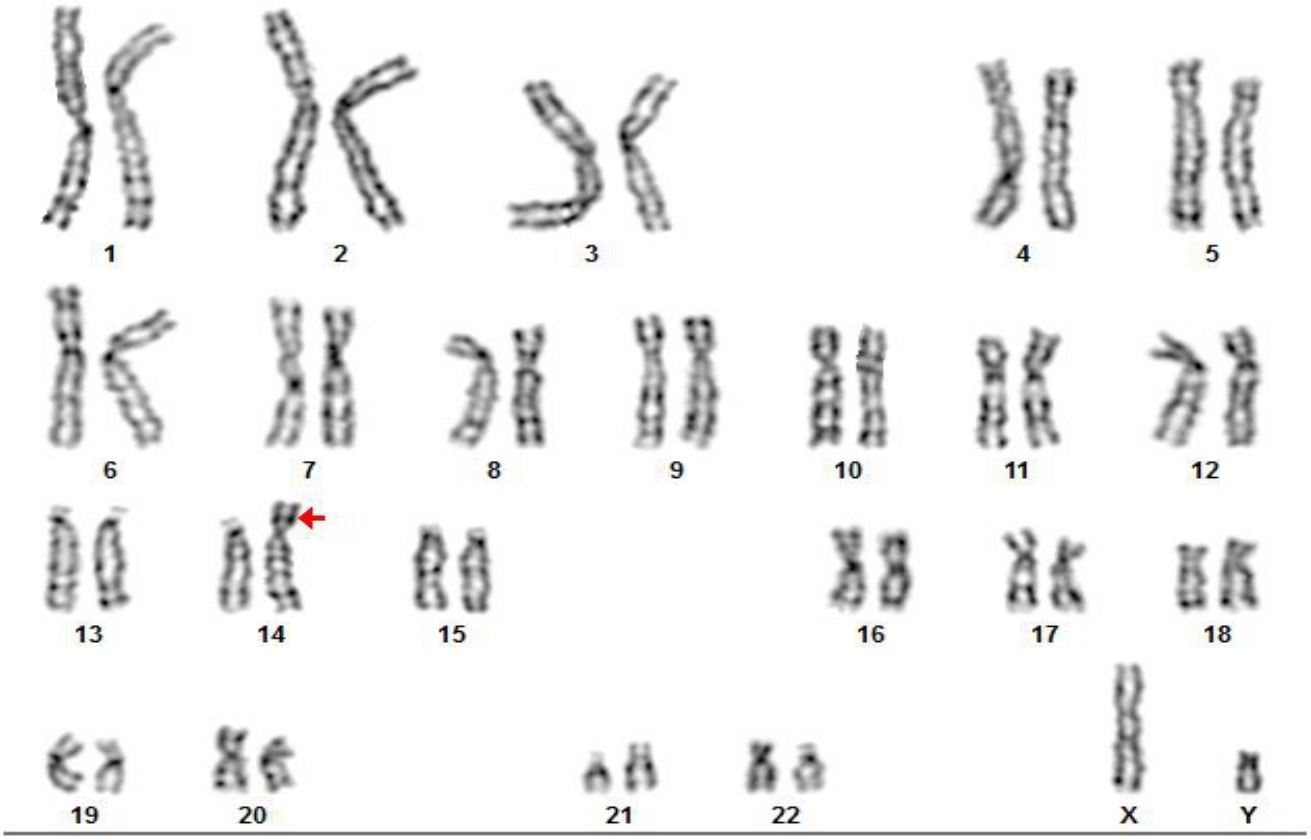
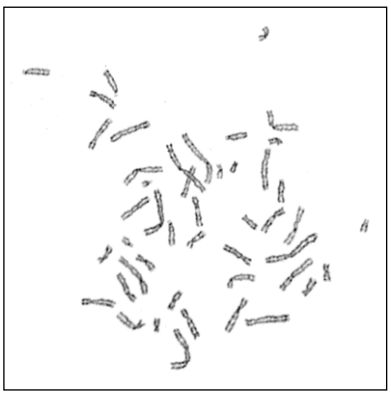
REPORT OF KARYOTYPE ANALYSIS



KARYOTYPE RESULT : **46,XY,der(14;21)(q10;q10),+21**

BAND RESOLUTION : **550**



Note:Results are interpreted on basis of all metaphases analyzed.This Karyotype is only a representation

-- End of Report --

MC-2139

Tests marked with NABL symbol are accredited by NABL vide Certificate no MC-2139

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