Mr. HARGUN SINGH AMBALA, CHEMBUR Tel No : 8607344999 PIN No: 133001 PID NO: P542000270047 Age: 8.0 Year(s) Sex: Male	Reference: Sample Collected At: Dr vinay kumar chopra Kos diagnostic lab, 6349/i, nicholson road, ambala cantt, hry 133001. PROCESSING LOCATION:- Metropolis Healthcare Ltd, Unit No. 409- 416, 4th Floor, Commercial Building-1, Kohinoor Mall, Mumbai-70	VID: 54203320008686 Registered On: 30/12/2020 01:18 PM Collected On: 01/01/2021 2:03PM Reported On: 13/01/2021 07:11 PM
Karyotyping by G-Banding Peripheral Blood		
INTERNAL LAB NO.	29-21-K (This report supersedes the previous report dated 12/01/2020 and is amended for age confirmation and doctor discussion.)	
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	47,XY,+21	
INTERPRETATION	Analysis revealed 45 autosomes and 2 sex chromosomes with presence of an extra copy of chromosome 21 in all the observed cells.	
COMMENTS	Report consistent with Down syndrome (Trisomy 21).	
RECOMMENDATION(S)	Kindly correlate clinically. 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. In case of any discrepancy kindly send new sample for confirmation. For any queries please feel free to contact at Department of Medical Genetics on 022-50560767.	

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.

**Dr. Jaya Vyas** PhD Applied Biology Sr Consultant, Medical Genetics

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Mr. Dhananjay Pathak M. Sc. Analyst, Medical Genetics, Metropolis - Mumbai

## Mr. HARGUN SINGH



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## **REPORT OF KARYOTYPE ANALYSIS** 3 13 14 16 17 18 15 22 19 20 21 v X **KARYOTYPE RESULT** : 47,XY,+21 **BAND RESOLUTION** : 550

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

-- End of Report --

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