Patient Name	:	Bill Date	:
DOB/Age/Gender	:	Sample Collected	:
Patient ID / UHID	:	Sample Received	:
Referred By	:	Report Date	:
Sample Type	:	Barcode No	:
Client	:	Report Status	:

CYTOGENETICS REPORT

Karyotyping: Blood Lympho Culture, Single

CLINICAL INDICATION Known Down's Syndrome

SUMMARY OF RESULTS **ABNORMAL FEMALE KARYOTYPE CONSISTENT WITH DOWN SYNDROME**

NOMENCLATURE **47,XX,+21**
(As per International System for Human Cytogenomic Nomenclature, ISCN,2020)

CLINICAL INTERPRETATION

An abnormal female chromosome complement with an additional chromosome 21 was observed in all metaphases. This result is consistent with the clinical diagnosis of Down syndrome.

Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. People with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory and hearing problems, Alzheimer's disease, childhood leukemia, and thyroid conditions. Many of these conditions are now treatable, so most people with Down syndrome lead healthy lives. Early treatment programs can help improve skills. They may include speech, physical, occupational, and/or educational therapy. With support and treatment, many people with Down syndrome live happy, productive lives. (http://www.ndss.org/Down-Syndrome/Down-Syndrome-Facts/ : NIH: National Institute of Child Health and Human Development).

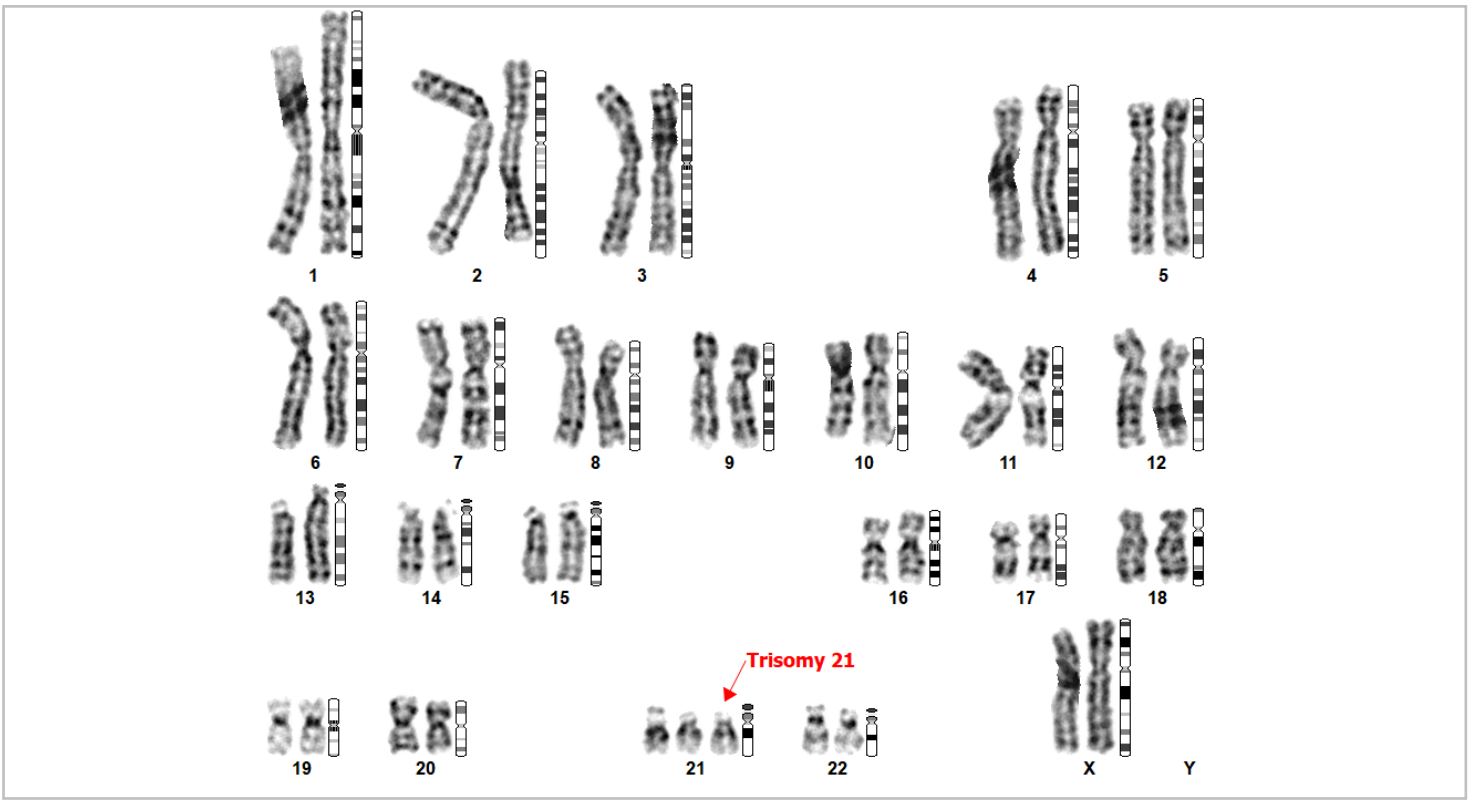
Within the limits of standard cytogenetic methodologies, the chromosomes of the patient showed abnormal G-banding patterns with evidence of aneuploidy or without apparent structural abnormality or rearrangement. The following possibilities, although rare, cannot be ruled out: a) low level mosaicism, b) very subtle rearrangements, c) genetic disorders that cannot be detected beyond the resolution of by standard cytogenetic methods.

RECOMMENDATION Genetic Counseling for the family is recommended.

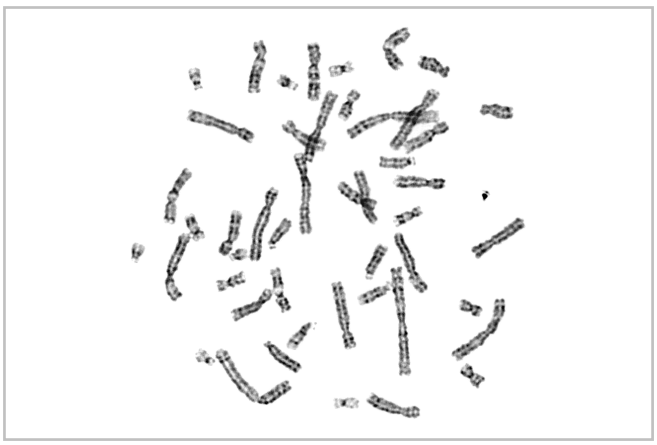
SAMPLE DESCRIPTION The sample was of optimal quality for conventional cytogenetics culture techniques. The 72 hours of stimulated peripheral blood sample was initiated in karyotyping medium yielded analyzable metaphases for karyotype.

Dr. Ashish Fauzdar
PhD (Genetics), AIIMS
Head of Clinical Genomics & Cytogenetics

KARYOTYPE IMAGE:



JAYSHREE SINGHA **Karyotype: 47,XX,+21** **Barcode No: CG002617**



METHOD:	G-BANDING
Metaphase Counted:	20
Metaphase Analyzed:	10
Metaphase Karyotyped:	10
Banding Resolution:	**625**
Metaphase Quality:	**Good**

Ms. Ritu (Jr. Scientist)
Cytogenetics

Dr. Ashish Fauzdar
PhD (Genetics), AIIMS
Head of Clinical Genomics

Reviewed and Signed out on: 14-Feb-2023

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1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

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