Name: KALIKA

Printed on

12-03-22

Age/Gender: 8 M/Female

Lab ID:

012202240039

# **CYTOGENETICS REPORT**

## Test Name:Karyotype - Blood

### CLINICAL INDICATION:

Monogoloid features

## **RESULTS:**

Method:	G-banding
Metaphases counted:	20
Metaphases analyzed:	20
Metaphases karyotyped:	20
Banding Resolution:	400
Karyotype (ISCN 2016):	47, XX,+21

#### INTERPRETATION:

This karyotype shows three copies of chromosome 21 (Trisomy 21) in all metaphases examined. Trisomy 21 is associated with features of Down syndrome. This finding is consistent with a clinical diagnosis of Down syndrome [MIM #190685]. Down syndrome is one of the most common chromosomal abnormalities in live born children and has a well-defined clinical presentation, including distinctive facial features and mild to moderate intellectual disability. Individuals with Down syndrome may also have congenital malformations of the heart and/or gastrointestinal tract and conductive hearing loss.

#### **RECOMMENDATION:**

Due to the occurrence of one child with Trisomy in this family, prenatal chromosome analysis should be offered for any future pregnancy of these parents. In addition, a genetic evaluation is recommended for this individual and genetic counseling is recommended for this family.

# CYTOGENETICS REPORT

### KARYOTYPE:

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**Disclaimer:** Although the methodology used in this analysis and interpretation is highly accurate, it does not detect small rearrangements and very low-level mosaicism, which are detectable only by molecular methods. Failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders.

\*\*\* End Of Report \*\*\*