NAME: Baby. ARISHA Accession No.: 111261 Age/Gender: 5 M/Female BC2400992 Specimen ID: Lab NO: 012410070031 Specimen: Whole Blood HEPARIN Referred BY: Dr. Vinay Chopra Collected: 07/Oct/2024 12:01PM Remark: 07/Oct/2024 11:59AM Registered: Reported: 26/Oct/2024 02:11PM

CYTOGENETICS REPORT

Test Name:Karyotype - Blood

CLINICAL INDICATION:

? Down's Syndrome

RESULT:

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.

Tara Nath Quality Manager

Mr. Brijesh Authorised Signatory PhD(P)

DR. S. KUMAR MBBS, MD Consultant Pathologist



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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. LABASSURE can assist the physician in determining the appropriate test in the context of clinical indications. This report has been reviewed and electronically signed by:

*** End Of Report ***

NOTE:- SAMPLE WAS OUTSOURCED.

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Tara Nath Quality Manager

KARYOTYPE:

Mr. Brijesh Authorised Signatory PhD(P)

DR. S. KUMAR MBBS, MD Consultant Pathologist

