

Booking Date 11/03/2022

Patient ID 012203110022

Printed on 08/04/2022

Name Nayra

Age 9 Months

Sex F

Ref By Aamit Enterprises

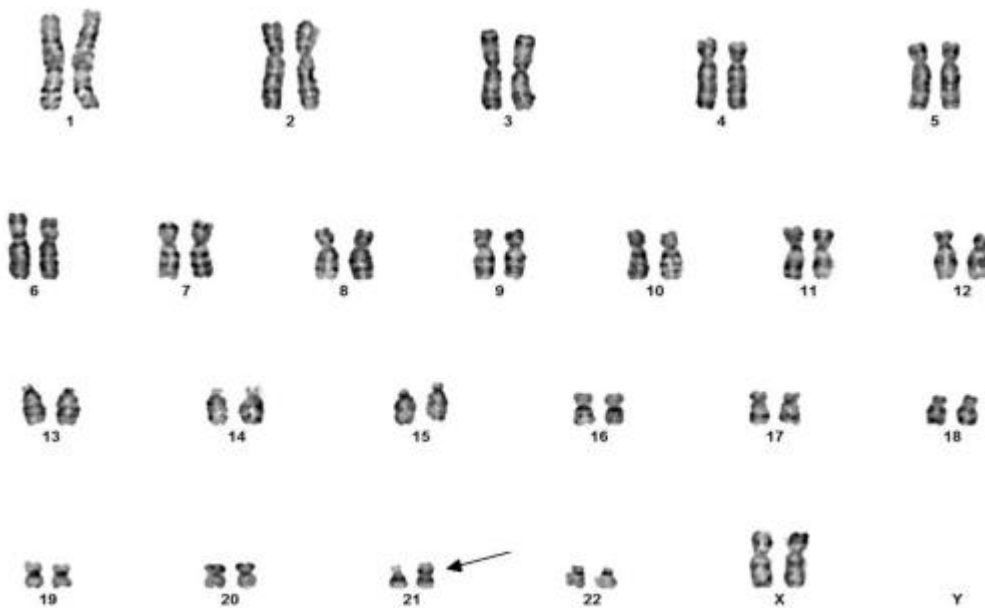
DIAGNOSIS: Down syndrome

KARYOTYPE: 47, XX,+21,der (21;21)(q10;q10)

INTERPRETATION: This karyotype shows a Robertsonian translocation involving two chromosomes 21, as well as a normal chromosome 21. The baby is thus trisomic for the long arm of 21, which is predictive of features 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. Parental chromosome studies are recommended to determine if either parent carries the balanced form of this 21;21 translocation.

KARYOTYPE:

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Peripheral Blood Chromosomal Analysis:

Please Note: 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.

For test performed on specimens received or collected from non-MolQ locations, it is presumed that the specimen belongs to the patient named or identified as labeled on the container/test request and such verification has been carried out at the point generation of the said specimen by the sender.

MolQ will be responsible only for the analytical part of test carried out. All other responsibility will be of referring Laboratory.