

Booking Date: 18/04/2024

Patient ID: 042404180013

Printed on: 09/05/2024

Name : Dawood Ahmad

Age: 5 Years

Sex: M

Ref By : GK Labs, Srinagar

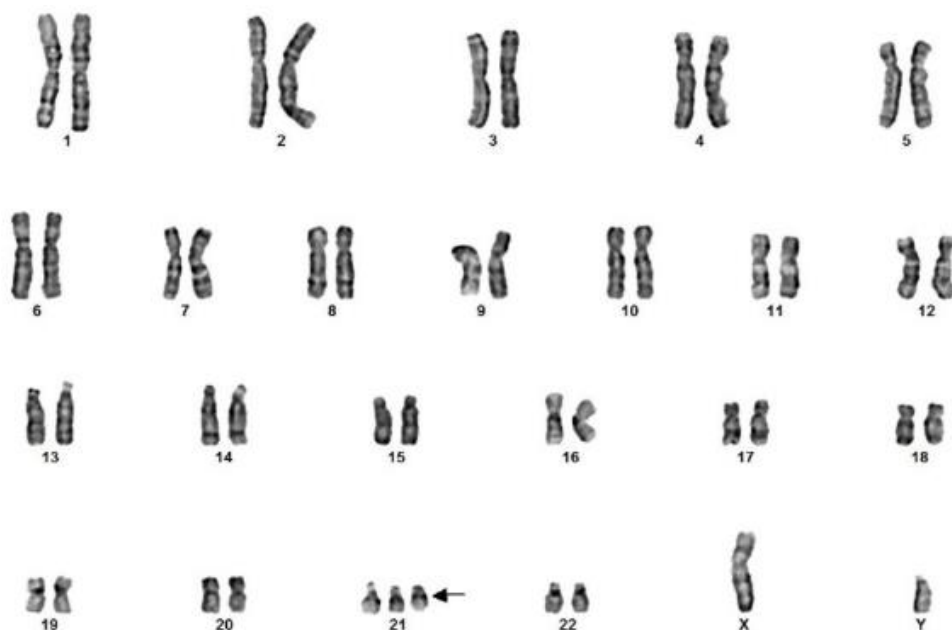
DIAGNOSIS: Karyotype – Down’s Syndrome

KARYOTYPE: 47, XY, +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.

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.

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.

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.