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Cytogenetic, Molecular and FISH Analysis of an Isodicentric Chromosome 21 idic(21)(q22.3) in a Mildly-Affected Patient with Down Syndrome

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ABSTRACT Most cases of Down syndrome (DS) result from a supernumerary marker chromosome 21; however there are rare cases in which DS is due to partial trisomy of chromosome 21, involving various segments of the chromosome. The characterization of DS that are due to partial trisomy 21 allows the phenotype to be correlated with the genotype. We present a case of a five-year-old male referred for cytogenetic analysis because of mild mental retardation and facial features typical of Down syndrome for whom karyotypic analysis showed a “mirror” reverse tandem duplication of chromosome 21. Fluorescence In-Situ Hybridization (FISH) using a half YAC containing the telomere of chromosome 21q and the adjacent marker D21S1575 revealed the deletion of both copies of this region on the translocated chromosome. Microsatellite analysis further delineated the breakpoint to be between CD18 and D21S1446, excluding the possibility of uniparental disomy and demonstrated that the rearranged chromosome was of paternal origin.