

Trisomy 9p Syndrome in a Mentally Retarded Female Inherited from Maternal Reciprocal Translocation

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ABSTRACT A rare case of trisomy 9p syndrome resulting from maternal reciprocal translocation is reported here, in a 14-year old female child with mental and motor developmental delay. The characteristic phenotype features of this syndrome such as dysmorphic features, congenital anomalies, severe mental retardation and gross delay in speech were observed. The cytogenetic study showed 47,XX, +der (9), (9pter→9q12; Xp22.2) mat. The mother was mentally normal, and her karyotype revealed chromosomal abnormality in terms of reciprocal translocation 46, XX, t (X; 9)(p22.2→ter; q13!ter), confirming her as a carrier and thus, leading to the inheritance of trisomy 9p in this girl child.