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DNA Diagnosis of Down Syndrome Using Polymerase Chain Reaction and Polymorphic Microsatellite Markers

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ABSTRACT For antenatal diagnosis of Down syndrome (trisomy 21) and for the determination of parental origin of nondisjoined chromosome, we have evaluated a simple PCR based DNA diagnostic method where polymorphic allelic fragments were resolved in 6% polyacrylamide gel. Three families, DS5, DS7 and DS9, each with a single child with trisomy 21 were studied, using two polymorphic microsatellite markers D21S2055 and D21S11 located on the long arm of the chromosome 21. Trisomy 21 was identified by the presence of three distinct alleles while the origin of nondisjunction was found to be maternal in all three families. The PCR based DNA diagnostic method was found to be sensitive, reproducible and most efficient not only for diagnosis of trisomy 21, but also for tracing allelic transmission from parents to the offspring.

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