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Prenatal Diagnosis in Pericentric Inversion 6

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ABSTRACT A four-year-old girl, the proband along with her mother in her subsequent pregnancy was referred to the genetic clinic for evaluation of global developmental delay with a normal karyotype study. On evaluation, dysmorphic features prompted to repeat the karyotype assessment. An unbalanced pericentric inversion of chromosome 6 in the index child was noticed. This was followed by identification of a balanced carrier status in the mother. Cytogenetic study using amniocentesis performed at our center for the subsequent pregnancy revealed balanced pericentric inversion of chromosome 6 in the fetus to that identified in the mother; a prenatal diagnosis was reached. The couple terminated the pregnancy following the diagnosis at 18 weeks.