Evaluation of Pregnancies in 25 Families with Balanced/Unbalanced Chromosomal Translocations

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ABSTRACT The researchers evaluated pregnancies in families with balanced/unbalanced translocations. This clinical cohort consisted of 25 pregnancies with balanced/unbalanced chromosomal translocations in family member(s) (maternal, paternal, fetal, abortion material, and/or previous fetus(es)) who underwent prenatal diagnosis. Translocations were observed in 18 cases (14 balanced and 4 unbalanced translocations). The researchers found 2 and 12 cases among the chromosomal translocations were paternal and maternal in origin, respectively. The researchers demonstrated that parent karyotypes were normal in 4 cases, while only maternal karyotypes were normal in 3 cases with unknown paternal karyotypes. Five of the prenatally diagnosed chromosomal abnormalities were Robertsonian and 13 were reciprocal translocations. Among the Robertsonian translocations, 2 were unbalanced. Early fetal loss or recurrent miscarriages were observed in previous history of 10(40%) and 6(24%) respectively. Prenatal diagnosis is critical in pregnancies with balanced/unbalanced chromosomal translocations in a member(s) of the family or those with poor gestational histories.