Adjacent 2 translocation involving 13q and 21q

A reciprocal translocation producing an adjacent 2 segregation is a relatively rare event. The study by Jalbert and Sele¹ described a small number of translocations falling into this category. The case we describe is, to our knowledge, the first involving chromosomes 13 and 21 with an adjacent 2 disjunction in the infant, and a balanced reciprocal translocation involving the long arms of chromosomes 13 and 21 in the mother.

The proband was the first child of young, healthy, non-consanguineous parents, born at 41 weeks' gestation. At delivery, oligohydramnios was noted. The infant's weight was 2480 g and head circumference 32 cm. On physical examination, the following abnormalities were noted: weak cry; sloping forehead and frontal upsweep; craniotabes with a third fontanelle; unruly hair pattern; bulbous nose with a septum extending below the nasal alae; ulnar deviation at the wrists; and wide big toes. Subsequently, congestive heart failure due to congenital heart disease appeared. The infant failed to respond to therapy for heart failure and died on the fifth postpartum day. Necropsy showed an endocardial cushion defect with common atria, common ventricles, common A-V valve, preductal coarctation of the aorta, and persistent ductus arteriosus.

Chromosome analysis was performed on metaphase and prophase cells cultured from a peripheral blood

sample from the proband using GTG banding. The proband's karyotype was 46,XX,-21,+der(13),t(13;21)(q14;q11)mat (figure a). The mother's karyotype was 46,XX,t(13;21)(q14;q11) (figure b). The infant's genotype is trisomic for nearly half the long arm of chromosome 13. This derivative portion also carries the 21q11 band. Moreover, the genotype is monosomic for the centromere-satellite region of chromosome 21.

The infant's genotype must have arisen through adjacent 2 disjunction, which is a relatively uncommon event. For example, in Jalbert and Sele's study, adjacent 2 disjunction was noted in only seven of the 161 recorded families.¹

Adjacent 2 translocations usually produce moderate imbalances compared to those arising from adjacent 1 and 3:1 segregations. The physical abnormalities, including oligohydramnios and low birthweight, noted in our proband do not present a phenotype consistent with the few other reported cases of trisomy for the proximal portion of the long arm of chromosome 13.^{2,3}

Since, by all reported cases, adjacent 1 and 3:1 do not occur in families where adjacent 2 occurs, it is possible that the fetus with the translocation products from these disjunctions may not survive, or some other factor(s) select against adjacent 1 and 3:1 segregation. Adjacent 2 disjunction may be repetitive in subsequent pregnancies. Moreover, it is possible that from the same adjacent 2 disjunction involving chromosomes 13 and 21, Down's syndrome genotype/phenotype could result. This segregation could also result in monosomy for a significant portion of the long arm of chromosome 13 and therefore might be lethal.

We have described here a new chromosome translocation that supports previous findings in families with adjacent translocations. It is important to recognise this

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FIGURE (a) Karyotype of the proband: 46,XX,-21,+der(13),t(13;21)(q14;q11)mat. (b) The reciprocal translocation, t(13;21)(q14;q11), in the proband's mother. Arrows point to the breakpoints on the ideograms corresponding to the chromosomes.

special class of translocations for both diagnosis and counselling.

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