Original Investigations

Reciprocal Translocation t(5;6)(p13;q27) Through Three Generations: Case Report of cri du chat Syndrome

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Summary. A male infant with cri du chat syndrome was found to have a deletion of the short arm of No.5 chromosome and which was due to maternal reciprocal translocation t(5;6)(p13;q27). His elder sister and his grandfather were also identified as the translocation carriers.

Introduction

Cri du chat syndrome was the first autosomal deletion syndrome which was reported by Lejeune et al. (1963). The clinical entity was well defined and over 300 cases have been reported. Niebuhr (1978) reviewed this syndrome and pointed out that more than 10% of this syndrome were due to the parental translocation. To date, over 50 cases of this syndrome (32 families) due to the parental (maternal; 25, paternal; 7) translocations were reported (de Grouchy and Gabilan, 1965; Back et al., 1978; Char, 1974; Carpentier et al., 1972; Lejeune et al., 1965; Mennichen et al., 1968; Oishi et al., 1973). We added the above a case of cri du chat syndrome due to maternal reciprocal translocation. The balanced translocation was confirmed to be inherited through three generations.

Cytogenetic Studies

Chromosome analyses were done from the peripheral blood of the propositus and his family. Routine G-banding method revealed the deletion of the short arm of No.5 chromosome of the propositus. The first pregnancy of the mother ended in spontaneous abortion in three months of gestation. The elder sister of the propositus was healthy. During the pregnancy of the propositus, no abnormalities were reported. Birth weight was 3100g (−0.25 S.D.), height 46.6 cm (−1.6 S.D.) and head circumference 32.7 cm (−0.53 S.D.). The external features of the baby were: a round face, microcephaly, micrognathia, almond-shaped eyes with bilateral epicanthal folds, downward slanting, hypertelorism, cleft lip at the right corner of the mouth, high arched palate, low-set and malformed ears with a preauricular appendix on the right side, short neck, simian crease in the left hand, and rigidity of the extremities. No heart murmur was noted. The chest X-ray, ECG, EEG, and brain echography were within normal range. He had characteristic mewing cry. Indirect laryngoscopy revealed no abnormalities. Laboratory findings were within normal range. The baby failed to thrive and was severe to gain weight. He died at 11 weeks of age because of respiratory difficulty. The autopsy was not allowed.

Case Report

The propositus, a male infant, was born in 1975 in the 42 weeks of gestation. The baby was the second child of healthy parents, the father 29 years old and the mother 27 years old. The first pregnancy of the mother ended in spontaneous abortion in three months of gestation. The elder sister of the propositus was healthy. During the pregnancy of the propositus, no abnormalities were reported. Birth weight was 3100g (−0.25 S.D.), height 46.6 cm (−1.6 S.D.) and head circumference 32.7 cm (−0.53 S.D.). The external features of the baby were: a round face, microcephaly, micrognathia, almond-shaped eyes with bilateral epicanthal folds, downward slanting, hypertelorism, cleft lip at the right corner of the mouth, high arched palate, low-set and malformed ears with a preauricular appendix on the right side, short neck, simian crease in the left hand, and rigidity of the extremities. No heart murmur was noted. The chest X-ray, ECG, EEG, and brain echography were within normal range. He had characteristic mewing cry. Indirect laryngoscopy revealed no abnormalities. Laboratory findings were within normal range. The baby failed to thrive and was severe to gain weight. He died at 11 weeks of age because of respiratory difficulty. The autopsy was not allowed.

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Fig. 1. Partial karyotype of the carrier mother. Derivative chromosomes are indicated by lines

- Derivative chromosomes
- Normal karyotype
- Balanced translocation carrier
- Normal, not karyotyped
- Spontaneous abortion
- Artificial abortion
- Dead
- Propositus
- Consanguinity

Fig. 2. Pedigree of the family with t(5;6) translocation

- A fetus (IV-4) with normal phenotype was aborted artificially in 27 weeks of gestation and found to have normal female karyotype. A girl (III-8) who died of enteritis at ten years of age, had been said to have mewing cry, round face, and mental-growth retardation which was so severe as she could not walk and speak until she died. In the process of this family studies, we had the information from other hospital that a male infant (IV-10) with typical cri du chat syndrome was revealed to have the same karyotype as our case. His mother (III-13) and his grandmother (II-15) were translocation carriers. Therefore, two translocation carriers were found in generation II. Five infants out of their nine children (II-9, 11, 12, 14, and 16) died in early age.

**Discussion**

The clinical features and the karyotypes of cri du chat syndrome due to the parental translocation which were reported with distinct exchange points are presented in Table 1. Our case would be the first report in which the translocation occurred between No. 5 and No. 6 chromosome. It might be one question whether cri du chat syndrome due to the parental reciprocal translocation has typical clinical features as those of classical cri du chat syndrome or not. The cri du chat syndrome due to the parental translocation has the deletion for the short arm of No. 5 chromosome with some amount of duplication at the same time, while the classical type of this syndrome has a simple deletion. In three cases of cri du chat syndrome, the clinical features were combined with those of partial 10p trisomy (Noel et al., 1976), 13 trisomy (Leisti et al., 1975), and partial 13q trisomy (Back et al., 1978). The case which was reported by Fried et al. (1976) had “atypical” clinical features of cri du chat syndrome. Our cases which had minute duplication for 6q27→6qter segment besides the deficiency for 5p13→5pter segment showed typical features of cri du chat syndrome. The duplicated chromosomal materials would be inactive or too little to have phenotypic effects. Trisomy 5p which may be derived from adjacent I segregation in gametogenesis of the translocation carrier could not be found in our pedigree.