An unusual chromosomal segregation in a family with a translocation between chromosomes 3 and 12

Summary. A family is reported in which two infants were born with different types of congenital abnormalities. Chromosome studies on one of the infants showed a partial trisomy of the short arms of a No. 3 chromosome. A family study showed many balanced translocation carriers who had extra chromosomal material on the long arms of a No. 12 chromosome.

Balanced translocation carriers for a chromosomal rearrangement have the potential for producing abnormal infants and are, therefore, adding to the abnormal biological load in our society. Because of the almost unlimited chromosomal rearrangements that can occur among chromosomes, it is to be expected that this form of chromosomal mutation will greatly contribute to the number of children that are born with congenital abnormalities.

We wish to report a family in which two infants were born with different types of congenital abnormalities. Both of these infants died within the first 6 months of life. Chromosomal studies on one of the infants revealed a chromosomal abnormality suggesting a partial trisomy of short arms of a chromosome 3. The second infant who died had a different complex of congenital anomalies and we assume that his anomalies were caused by a deletion of a No. 3 chromosome.

Case reports

III.5 was a female infant weighing 1520 g. The anterior and posterior fontanelles were longer than normal. The ears were low set and she had micrognathia. The peritoneum was incompletely closed. There was a bicornuate uterus. Both large toes were displaced medially. The child lived 36 hours. The immediate cause of death was atelectasis of the lungs. The infant was born at another hospital and no chromosomal studies were done.

III.8 was a male infant born after 37 weeks' gestation and weighed 1700 g at birth. The patient's facial appearance was unusual and he was slightly jaundiced shortly after birth. The child lived for approximately 6 months and died of a congenital heart condition. Chromosomal studies which had been done at another hospital revealed that the child had 46 chromosomes with a karyotype which showed extra chromosomal material on the long arms of a No. 12 chromosome.

At necropsy examination, he had an enlarged heart with stenosis of the mitral valve. There was a deformity of the coccyx associated with a pilonidal sinus. There were congenital deformities of the lungs and kidneys. The penis was short and deformed. The testes were undescended.

Chromosomal studies

Chromosomal studies were done on both parents and their five living children (Fig. 1). The chromosomes were stained using the usual Giemsa staining technique. In addition, they were also examined by staining for G-banding and fluorescence (Fig. 2). The father's chromosomes were normal. The mother and all five children had 46 chromosomes which on karyotype analysis showed the same chromosomal rearrangement. There was a partial deletion of the short arms of a No. 3 chromosome and extra chromosomal material on the terminal end of the long arms of one of the No. 12 chromosomes (Figs. 2 and 3).
Case reports

Discussion

This pedigree demonstrates a reciprocal balanced translocation in seven family members, II.2, II.4, III.1, III.2, III.3, III.4, and III.7 (Fig. 3). An eighth individual in generation I undoubtedly had the same balanced translocation but, unfortunately, the individuals in generation I were not available for chromosomal testing. The unbalanced form of the translocation was definitely identified in an infant born with multiple congenital anomalies (III.8). Infant III.5, with a different complexity of congenital anomalies, died shortly after birth without having had chromosomal studies done.

This family represents an unusual segregation of a reciprocal translocation. In generations II and III there was a total of 11 pregnancies in which only balanced and unbalanced forms of the translocation are represented. There are no members representing the complementary normal karyotype. The two

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Fig. 1. Family pedigree.

Fig. 2. Fluorescing chromosomes showing extra chromosomal material on the long arms of a No. 12 chromosome and a deletion of chromosomal material on the short arms of a No. 3 chromosome.
spontaneous abortions (II.3 and III.6) were not tested for their chromosomal patterns. 

The chromosomal exchange in this family occurred between the short arms of a No. 3 chromosome and the long arms of No. 12 chromosome (Figs. 1 and 2). There have been other examples of a reciprocal translocation between a No. 3 chromosome and another chromosome (Clarke et al, 1964; Falek, Schmidt, and Jervis, 1966; Smith, Shear, and Jalowayski, 1969; Soukup et al, 1969). In the family reported by Falek et al (1966), there was an apparent translocation between chromosomes 3 and 21 or 22, with the unbalanced patients having an additional segment from one end of a No. 3 chromosome. In this family, the unbalanced patients were clinically diagnosed as having the de Lange syndrome.

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