74. A Case of Partial Trisomy 4q Resulting from Familial (4;9)(q23;p24) Translocation

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Current chromosome banding techniques have made possible the identification and the analysis of structural aberrations of chromosomes to a great extent. Current literature refers to 34 cases of partial trisomy 4q in association with a variety of clinical features. A female infant with multiple congenital malformations associated with partial trisomy 4q (q23→qter) resulted from maternal balanced (4;9) translocation is reported in this paper.

Case reports. The proband is a female infant born as the first child between a 26-year-old mother and a 31-year-old father. Both parents are phenotypically normal and not consanguineous and there was no evidence for atomic bomb exposure. They have no history of spontaneous abortions and stillbirth.

The proband was 3130 g in birth weight at 39 weeks of gestation. Her remarkable clinical signs were hypotonia, brachycephaly, flat occiput, broad nasal bridge, hypertelorism, blepharophimosis of right side, slightly downturned corners of mouth, dysplasia helix, low set and posterior rotated ears, short and webbed neck, congenital heart disease, wide-set nipple, and hypoplasia of metacarpal bones at bilateral thumbs (Fig. 1).

Fig. 1. Clinical features of the proband.

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Cytological findings. The chromosomes were investigated in well-delineated cells from leucocyte cultures established from the propositus and her parents. Their chromosomes were analysed following the G- and Q-banding procedures.

The father's karyotype was normal. The mother's cells were 46 in chromosome number, but one of no. 4 chromosomes was deleted at q23. The missing segment of this chromosome was translocated on the terminal portion of the short arm of one no. 9 chromosome. Thus the mother's karyotype was characterized by a reciprocal translocation between no. 4 and no. 9 as represented by 46,XX,t(4;9)(q23;p24) (Fig. 2A).

Fig. 2. G-banding partial karyotype of the mother (A) showing a reciprocal translocation between no. 4 and no. 9 chromosome: 46,XX,t(4;9)(q23;p24), and the propositus (B) having the trisomic segment of no. 4 on no. 9: 46,XX,der(9),t(4;9)(q23;p24) mat.

The karyotype of the patient had two normal no. 4 chromosomes and one normal no. 9 chromosome. The mate of the no. 9 showed the same type of translocation t(4;9) as that observed in the mother.

The karyotype of the patient is represented by 46,XX,der(9),t(4;9)(q23;p24) mat (Fig. 2B).

Remarks and conclusion. Following the first information by Hoehn *et al.* (1971), 34 cases of partial trisomy 4q are referred to in the literature: 26 cases were summarized by Bonfante *et al.* (1979), and the remaining 8 were reported by Oka *et al.* (1978), Andrie *et al.* (1979), Kelly *et al.* (1979), Mattei *et al.* (1979), Adachi and Sano (1979), Fryns and Van den Berghe (1980), and Stoll and Roth (1980). It was shown that the trisomic segment of no. 4 varied from 4q21 to 4qter and that this caused variable phenotypic clinical features, corresponding to the variation of the length of the trisomic segment 4q.

Clinical features common to all reported cases were summarized by Oka *et al.* (1978), as follows: Psychomotor retardation, abnormalities of ocular and/or palpebral fissures, low set and/or malformed ears, undescended testis in males and low birth weight, microcephaly, hypertelorism, malformed digits and toes, depressed on wide nose bridge, congenital heart disease, abnormalities of urogenital system, and hypo- or hypertonia. The present case had most of these clinical signs, except low birth weight, microcephaly, malformed toes, and abnormalities of urogenital system. The mother of the proband is most probably a carrier of a balanced translocation t(4;9) as already mentioned. The propositus could be accounted for by the gametic segregation of the abnormal chromosome no. 9 resulting in partial trisomy for the long arm of the no. 4 (q23→qter).

It is then concluded that the propositus possesses the karyotype, 46,XX,der(9),t(4;9)(q23;p24)mat, and her mother is a carrier of a balanced translocation, 46,XX,der(9),t(4;9)(q23;p24).

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