**120. Partial Trisomy 3q22 Involving the No. 15 Short
Arm without Association with Cornelia
de Lange Syndrome**

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Current banding chromosome analyses have rendered possible to reveal partial trisomy translocated on the other chromosome.

Previously we had an opportunity to report a case with a long arm duplication of chromosome no. 3, sporadic in occurrence (Kadotani *et al.* 1979). In the present article is reported a new case involving an abnormal chromosome no. 15 which carries a translocated portion of the long arm (3q22). Conclusively the aberration here concerned was defined as partial trisomy 3q of maternal origin.

Case reports. The propositus was a one-year and two-month-old female baby, 8 kg in weight, with circumferences of 41 cm in head, and 71 cm in length. She was born to a 33-year-old mother and a 34-year-old father as a second child. There was no history of abortion, still birth, congenital malformations, and consanguinity in this couple. Her mother was one of the atomic bomb survivors exposed in Hiroshima. Clinical examinations revealed that her parents and elder sister were phenotypically normal. The birth weight of this patient was 2820 g at 41 weeks gestation. The growth retardation as well as physical and mental development of this child were specially outstanding (Fig. 1). Remarkable clinical signs of this child were as follows: Brachycephaly, short neck, round face, hypertelorism, inverted epicanthus, congenital glaucoma at both eyes, saddle nose, high arched palate, wide-set nipples, hyperextensible hip in both sides, disturbance of gaits due to underdevelopment of the motor nerve, and mental deficiency.

Cytological findings. Chromosome slides for study were prepared from leucocyte cultures established from the propositus and her parents. For differential staining, the G- and Q-banding procedures

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were applied to the slides made by the air-drying procedure. Chromosome counts were done with 50 well-delineated metaphases in each. Karyotypes were analysed in 20 cells each from conventional, G- and Q-banding preparations.

Fig. 1. A-C: External pictures of the proband.

The karyotype of the proband derived from the conventional Giemsa specimens showed 46 chromosomes in which a no. 15 chromosome had an unusually elongated satellite on its short arm. The assessment of the unusual element became possible by the examinations of the maternal karyotype. The results of the examinations in the parents indicated that the mother was the balanced carrier of insertion of a long arm segment of no. 3 into the terminal portion of the satellite of no. 15 chromosome. The G- and Q-banding analyses made it possible to reveal that the segment q22 of chromosome no. 3 was missing, and that the missing segment was inserted into the terminal portion of the satellite of chromosome no. 15. Then, the chromosomal formula of the mother was designated as 46,XX,ins(15;3)(p13;q22) (Figs. 2, 3).

On the basis of the findings of the maternal translocation, it became evident that the aberration occurring in the proband corresponds to partial trisomy for the long arm of no. 3. Then, her chromosomal formula was represented by 46,XX,der(15), ins(15;3)(p13;q22)mat (Fig. 4).

The father and the elder sister of the proband were apparently normal in their karyotypes without showing any detectable chromosome change.

Remarks and conclusion. Several cases of partial trisomy shown by 3q have been announced with the discussion involving the correlation to Cornelia de Lange syndrome (Allderdice *et al.* 1975; Boue *et al.* 1974; Centerwall *et al.* 1977; Chiyo *et al.* 1976; Falek *et al.* 1966;

Fig. 2. G-(A) and partial Q-banding karyotype (B) of the mother, showing 46,XX,ins(15;3)(p13;q22). Balanced condition.

Fig. 3. Diagrams showing insertion of no. 3 long arm into the terminal portion of the satellite of chromosome 15. Balanced condition in mother.

Fear and Briggs 1979; Fineman *et al.* 1978; Fryns *et al.* 1978; Kadotani *et al.* 1979; Kondo *et al.* 1979; Mulcahy *et al.* 1979; Patil *et al.*

Fig. 4. G-(A) and partial Q-banding karyotype (B) of the proband showing 46,XX,der(15),ins(15;3)(p13;q22)mat.

1978; Salazar *et al.* 1978; Sciorra *et al.* 1977; Schwanitz *et al.* 1977; Sod *et al.* 1978; Stengel-Rutkowski *et al.* 1979; Wilson *et al.* 1978; Yunis *et al.* 1979). It has been shown that the clinical pictures of the trisomic anomalies of 3q are generally associated with the size and site of the trisomic segment.

The cases of trisomy of 3q described prior to our case were remarkable by having a relatively long segment of the long arm of no. 3. In contrast, the 3q of our case corresponded to a single short segment of no. 3, or 3q22.

The major clinical features of this case were mental retardation, glaucoma, inverted epicanthus and brachycephaly, without showing the signs of typical Cornelia de Lange syndrome. The mother of the proband is most probably a carrier of the balanced translocation involving the middle segment of a long arm of the no. 3 (q22) and the short arm of the no. 15 (Figs. 2, 3). She had one normal girl. The chromosome aberration of the proband could be accounted for as the result of the gametic segregation of the abnormal no. 15 chromosome, leading to a partially trisomic for the long arm of a no. 3 chromosome.

The conclusion is then possible that the propositus is characterized by the karyotype, 46,XX,der(15),ins(15;3)(p13;q22)mat, and that her mother is a carrier of the balanced translocation, 46,XX,ins(15;3)(p13;q22). This aberration is the first to be reported in man.

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