17. A Case with Partial Monosomy 10p

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Current banding chromosome analyses have rendered it possible to detect the chromosome number and various aberrations.

Currently six cases associated with genuine partial monosomy for the short arm of a chromosome no. 10 are referred to in the literature (Francke *et al.*, 1975; Shokier *et al.*, 1975; Berger *et al.*, 1977; Prieto *et al.*, 1978; Bourrouillou *et al.*, 1981; Fryns *et al.*, 1981). Because of rather rare occurrence of the cases, their clinical features have not been fully established at present. The present article describes some cytogenetic profiles of a boy with the above chromosome anomaly.

Case reports. The propositus was an eighteen-year-old boy, 58 kg in weight, with circumferences of 57 cm in the head and 84 cm in the breast, and 149 cm in length. He was born to a 38-year-old father and a 31-year-old mother as a third child. There was no history of abortion, still birth, congenital malformation, consanguinity and exposure to atomic bomb in this couple. Clinical examinations revealed that his parents, and older two brothers were phenotypically normal without evidence of mental deficiency. The birth weight of this patient was 3600 g at 42 weeks of gestation. Remarkable clinical signs of this patient were small of the head, flat occiput, trigono- and macrocephaly, antimongoloid slant, bilateral epicanthus, hypotelorism, hypertelorism, wide nasal bridge, thin upper-lip, high-arched palate, low set and small ears with adherent lobulus, wide-spaced nipples, and undescended testes, but there were no abnormalities in the hand, the feet, and the heart. He is deeply mentally retarded and I.Q. tested at 17 years of age was found to be 19.

Cytological findings. Chromosome preparations were made from the leucocyte culture established from the propositus. For the differential staining, the G-banding procedure was applied to air-dried slides prepared from the above subject. The chromosome count was done in 36 well-delineated metaphases. Karyotypes were analysed with 24 cells in each cell according to the conventional Giemsa and G-banding techniques.

Fig. 1. External profile of the proband.

Fig. 2. Diagram showing the position of the deletion of chromosome no. 10.

Fig. 3. G-banding karyotype of the proband, showing 46,XX,del(10)(p14→pter) with G-banding technique.

The karyotype of the proband derived from the conventional Giemsa specimens showed 46 chromosomes including a chromosome no. 10 having an unusually deleted short arm. On the basis of differential G-banding analysis in the proband, it was revealed that the abnormal short arm of the chromosome no. 10 resulted from the deletion of a part of the short arm of the chromosome no. 10 (p14→pter) (Figs. 2, 3).

Remarks and conclusion. Since the technique of banding analysis of the chromosomes had been established, the short arm deletion of the chromosome no. 10 has been reported in six cases up to the present time (Francke *et al.*, 1975; Shokier *et al.*, 1975; Berger *et al.*, 1977; Prieto *et al.*, 1978; Bourrouillou *et al.*, 1981; Fryns *et al.*, 1981). In the first four reports a terminal deletion of 10p with the break point at 10p13 was described; in the patient of Fryns *et al.* (1981) and ours 10p terminal deletion with the break point is located at 10p14. The clinical signs of the patients with the chromosome 10p— are in common: They are mental and growth retardation, occiput flat, macro- and trigonocephaly, antimongoloid slant, epicanthus, ptosis, hypotelorism, wide nasal bridge, high-arched palate, small low-set ears, heart anomalies, wide-spaced nipple, minor abnormalities of extremities, and undescended testis. Our case showed no abnormalities in extremities and heart. Further detailed chromosome analysis is necessary to provide further knowledge for such a partial monosomy of the short arm deletion of the chromosome no. 10.

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