18. A Case of Trisomy 8 Mosaicism

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Since the identification of C group chromosomes has become possible by the introduction of chromosome banding techniques, several cases of the trisomy 8 mosaicism have been contributed. We chanced to study one case of the trisomy 8 mosaicism. In this paper, the authors aimed to provide the major clinical and cytogenetic data of an additional case with trisomy 8 mosaicism.

Case reports. The propositus is a male infant born as the first child between a 23-year-old mother and a 28-year-old father. His parents are phenotypically normal and not consanguineous, and there was no evidence for the atomic bomb exposure. They have no history of spontaneous abortion and stillbirth.

The propositus was 2820 g in birth at 40 week gestation. His remarkable clinical signs were sparse hair, macrocephaly, hypertelorism, mild epicanthus, low-set ears with a folded cartilage-deficient pinna and distorted concha, broad bulbous and upturned nose, high arched palate, congenital heart anomaly (V.S.D.), palmar and planter deep skin furrows, short fifth fingers with clinodactyly and camptodactyly, undescended testis at right side, talipes equinovarus, and deformed- and flexed-toes. Computed tomography revealed that his brain was a genesis of corpus callosum (Fig. 2).

Cytological findings. Chromosome slides for study were prepared from leucocyte cultures established from the propositus and his parents. For differential staining, the G-banding procedures were applied to the slides after the air drying procedure. Chromosome counts were done with 111 well-delineated metaphases in the proband. Karyotypes were analysed on 27 cells based on conventional Giemsa and G-banding preparations. In sixty six (59.5%) out of 111 cells, one of the extra chromosome in the C group chromosome was counted. The extra C chromosome was identified as one of the no. 8 as a result of G-banding analysis. Forty five out of 111 cells (40.5%) so far examined, showed a normal male karyotype (46,XY) (Table I). Because of the presence of the normal cells, the occurrence of mosaicism

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Fig. 1. External profile of the proband.

Fig. 2. Computed tomography of the proband's brain.

is to be suggested in our case. The karyotypes of his parents were normal, based on 24 cells karyotyped in each.

Remarks and conclusion. Since the first report by de Grouchy *et al.* (1971), several cases of trisomy 8 mosaicism have been reported. In Japan, three cases were reported by Kondo (1976), Mino *et al.* (1976), and Shiraishi (1976). The clinical features of trisomy

Table I. Cytogenetic findings in the leucocyte cultures of the propositus

Karyotype	Total cells counted (%)	Total cells karyotyped
46,XY	45 (40.5)	5
47,XY,+8	66 (59.5)	12
Total	111 (100)	17

Fig. 3. G-banding karyotype of the propositus having a trisomy no. 8 chromosome: 47,XY,+8.

8 mosaicism were summarized by Rodewald *et al.* (1977). The common clinical manifestations are mild to moderate mental retardation, strabismus, osseous and soft tissue abnormalities, skull deformity, low-set and/or malformed ears, broad bulbous, upturned nose, and palmar and/or plantar deep skin furrows, short fifth fingers with clinodactyly and comptodactyly, abnormalities of hand and/or feet, restricted articular function, long slender trunk and a slender pelvis, and so on. On the other hand, each of Caspersson *et al.* (1972), Jehan *et al.* (1979), and Chandley *et al.* (1980) reported a case of trisomy 8 mosaicism with normal intellectual and physical development.

In a review of the literature, it was shown that the case of the trisomy 8 mosaicism shows considerable variations in the clinical manifestations with wide range of clinical findings. From the consideration on considerable variations in the clinical features of the trisomy 8 mosaicism, it seems to be influenced by the proportion of abnormal cells in the malformed sites. Thus, further investigations of trisomy 8 mosaicism patients are needed before the trisomy 8 mosaicism syndrome can be established.

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