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A CASE OF PARTIAL TRISOMY 22 RESULTING FROM MATERNAL 11/22 TRANSLOCATION

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Summary A malformed female infant with psychomotor and growth retardation was found to have partial trisomy 22 due to maternal translocation. The karyotype was designated as 47,XX,+der(22), t(11;22) (q25; q13)mat. Comparison was made with 14 other reported cases of partial trisomy 22.

INTRODUCTION

The development of banding techniques has led to the establishment of various new chromosome syndromes. Trisomy 22 was suspected by Uchida *et al.* (1968), and recognized as a clinical entity by Hsu *et al.* (1971) and Penchaszadeh and Cocco (1975).

In the present paper, a female infant of partial trisomy 22 resulting from maternal 11/22 translocation is described. The patient showed most of the cardinal features of the trisomy 22 syndrome.

CASE REPORT

The patient was born at term as the second child to unrelated parents. The parents and elder sib were phenotypically normal. No family history indicating mental retardation and congenital anomalies was present. The mother had experienced first-trimester miscarriages twice. At the child's birth, the mother and father were 25 and 30 years old, respectively. The pregnancy and delivery were uneventful, apart from mild asphyxia at birth (Apgar scores 7 at 1 min). The birth weight was 2,600 g, length 47.0 cm, and head circumference 32.0 cm. Twenty days after birth, the infant was referred to the Okayama Saiseikai Sougou Hospital for abnormal appearance and poor weight gain. Physically, the following abnormalities were noted (Fig. 1): microcephaly, bilateral blepharoptoses and short palpebral fissures, large and low-set ears with a right preauricular skin tag and bilateral sinuses, long and prominent philtrum, micrognathia, cleft palate, heart

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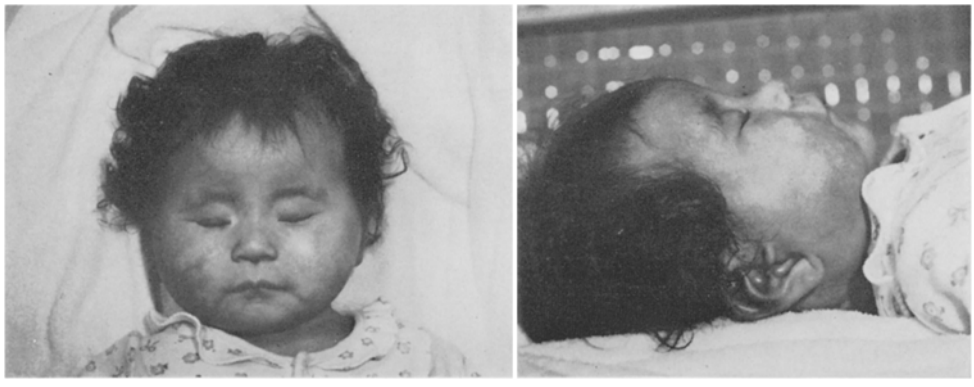

Fig. 1. Appearance of the patient (*post mortem*). Note large low-set ear, preauricular skin tag and sinus, long and prominent philtrum, and micrognathia.

murmurs, limited abduction of hip joints, and long slender fingers. Laboratory investigations including complete blood count, urinalysis, blood chemistry and chest roentgenogram were all within normal ranges. Electroencephalography demonstrated diffuse dysrrhythmia with increased slow waves. Dermatoglyphic study showed finger patterns consisting of 6 ulnar loops, 3 arches, and one radial loop on an unusual site (right fifth finger), a low total finger ridge count (36), absence of digital triradius b on both sides, and presence of an ulnar loop pattern on the right hypothenar area.

Her growth and psychomotor development was retarded. She began to smile at 4, and held her head at 5 months of age. At 6 months, the body weight was 6,180 g, length 61.0 cm, and head circumference 39.0 cm; all the figures were below the 3rd percentile. At 8 months, the infant died suddenly with high fever and convulsion. Autopsy showed atrial septal defect.

CYTOGENETIC FINDINGS

Cytogenetic studies employing GTG and RHG bandings were performed on cultured lymphocytes from the patient and her parents. In all cells of the patient was present an extra acrocentric chromosome, which was smaller than a G group chromosome and was frequently involved in satellite associations with D or G group chromosomes. The banded karyotype of the father was normal. That of the mother, on the other hand, showed a balanced translocation of the distal half of the long arm of chromosome 22 to the long arm of chromosome 11: t(11;22) (q25;q13). The extra chromosome of the patient seemed to be identical to the translocated chromosome 22 of the mother (Fig. 2). The karyotype of the patient, therefore, was designated as 47,XX,+der(22), t(11;22) (q25;q13)mat. The gain of the translocated chromosome 22 in the patient was considered to be the result of 3 : 1 segregation in meiosis I in the mother.

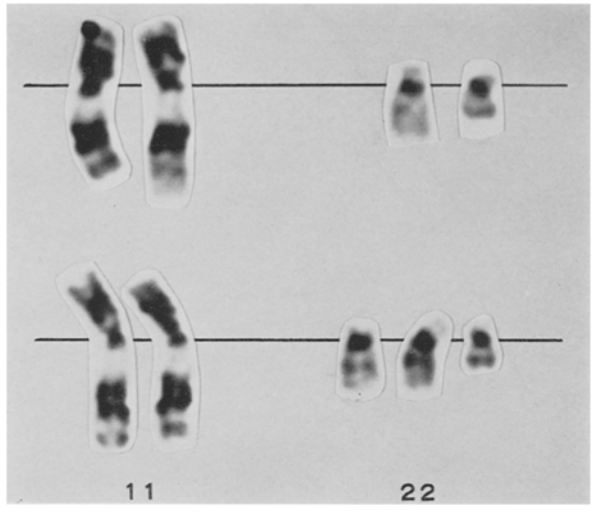

Fig. 2. Partial karyotypes of the patient and mother (G-banding).

DISCUSSION

Since the advent of banding techniques, 14 cases of partial trisomy 22 have been reported. The trisomic segment in these cases ranged from 22pter to 22q11 (McIntyre *et al.*, 1975), to 22q12 (Garlinger *et al.*, 1977), or to 22q13 (Alfi *et al.*, 1975; Matsui *et al.*, 1976; Bofinger and Soukup, 1977; Nakai *et al.*, 1977; Taillemite *et al.*, 1977; Feldman and Sparkes, 1978; Kadotani *et al.*, 1978). In five cases (Bühler *et al.*, 1972; Punnett *et al.*, 1973; Borgaonkar *et al.*, 1975; Zellweger *et al.*, 1976), however, break points were not determined. Parental translocations were identified in six cases (Borgaonkar *et al.*, 1973; McIntyre *et al.*, 1975; Bofinger and Soukup, 1977; Garlinger *et al.*, 1977; Nakai *et al.*, 1977; Feldman and Sparkes, 1978), and suspected in five cases (Bühler *et al.*, 1972; Punnett *et al.*, 1973; Alfi *et al.*, 1975; Zellweger *et al.*, 1976).

Clinical features of the cases of partial trisomy 22 were summarized in Table 1. For comparison, those of cases of full trisomy 22 identified by banding techniques were also listed. Although varied phenotypic expressions were noted in partial trisomy 22, several features were common. They included normal birth weight, mental and growth retardation, congenital heart disease, an antimongoloid slant of the palpebral fissures, preauricular skin tags and/or sinuses, large and/or low-set ears, micrognathia, congenital dislocated hips, strabismus, and hypotonia. On the other hand, microcephaly, beaked or bulbous nose, long philtrum, and finger abnormalities were less prominent in partial trisomy 22 as compared with full trisomy 22. The present case showed most of these common features, and additionally exhibited short palpebral fissures, blepharoptosis, long and prominent philtrum,

Table 1. Summarized clinical features of partial and full trisomy 22.

	Cases of partial trisomy 22	Cases of full trisomy 22[a]	Present case
Birth weight more than 2,500 g	11/11	13/25	+
Mental retardation	14/14	22/22	+
Growth retardation	11/14	20/22	+
Low-set and/or large ears	11/14	21/25	+
Preauricular tags and/or sinuses	10/14	18/25	+
Antimongoloid slant of the palpebral fissures	10/14	14/22	−
Congenital heart disease	10/14	20/26	+
Micrognathia	8/13	20/25	+
Congenital dislocated hip	8/13	8/22	+
Strabismus	7/11	12/19	−
Hypotonia	7/11	14/21	−
Microcephaly	7/13	17/21	+
Hypertelorism	5/9	6/12	−
Cleft palate or high-arched palate	6/13	14/26	+
Finger-like thumbs and/or long slender fingers	3/11	16/22	+
Beaked or bulbous nose	3/11	16/22	−
Imperforate anus	3/12	3/26	−
Craniofacial asymmetry	2/7	6/9	−
Long philtrum	1/11	16/23	+
Coloboma of iris	1/12	2/25	−

[a] References: Bass *et al.*, 1973; Begleiter *et al.*, 1976; Cervenka *et al.*, 1977; Emanuel *et al.*, 1976; Goodman *et al.*, 1971; Gustavson *et al.*, 1972; Hirschhorn *et al.*, 1973; Hsu *et al.*, 1971; Iselius and Faxelius, 1978; Lalchev *et al.*, 1978; Mollica *et al.*, 1977; Penchaszadeh and Cocco, 1975; Pérez-Castillo *et al.*, 1975; Punnett *et al.*, 1973; Shokeir, 1978; Uchida *et al.*, 1968, 1976; Vianello and Bonioli, 1975; Welter *et al.*, 1978.

long slender fingers and unusual dermatoglyphics. The phenotype of the present case was rather consistent with the trisomy 22 syndrome.

In view of the phenotypic similarity between cases of partial trisomy 22 Garlinger *et al.* (1977) have suggested a specific partial trisomy 22 syndrome, while Feldman and Sparkes (1978) have emphasized its phenotypic variability and indicated that phenotypically partial trisomy 22 was hard to separate from the reported cases of ful trisomy 22. In our compilation of cases of partial trisomy 22, the phenotypic variability was also apparent. It seems likely that trisomy for the short and proximal long arms of chromosome 22 is primarily responsible for the partial trisomy 22 syndrome. As indicated by Feldman and Sparkes (1978), the phenotypic variability of partial trisomy 22 may be explained either by the variable factors, such as amount or type of genetic materials which are retained on the distal long arm of chromosome 22, or by the different second autosome materials translocated on the supernumerary

chromosome 22. To clarify this problem, further observations of partial trisomy 22 using high-resolution banding techniques are required.

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