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A CASE OF PARTIAL 14 TRISOMY, 47,XX,+der(14),t(9p+;14q−)

Hanako TADA,[1] Tamiko SHINOHARA,[2] and Hisatoshi MIYATA[2]

[1]*Department of Pediatrics, Juntendo University School of Medicine, Tokyo 113, Japan*
[2]*Department of Human Cytogenetics, Japan Red Cross Medical Center, Tokyo 150, Japan*

Summary A malformed female infant had chromosome abnormality 47,XX,+der(14),t(9p+;14q−)mat. She had mental and growth retardation, craniofacial dysmorphy and hydrocephalus.

INTRODUCTION

About twenty cases of partial trisomy 14 have been reported to date, most of them being unbalanced offspring of translocation carriers. Clinical features seem to be influenced by the region of the trisomic segment of No. 14 chromosome and/or additional trisomic or monosomic segment of the other chromosome.

In this report, a female infant with a supernumerary marker chromosome is described, which is composed of the short arm, centromeric region and proximal part of the long arm of chromosome 14, and the distal segment of the short arm of No. 9 chromosome. The patient was a segregant of a carrier mother through 3 : 1 disjunction.

This case had already been reported orally in 1977 (Kataoka *et al.*, 1977).

CASE REPORT

The proband was a one-year-old female born to a thirty year-old mother after 43 weeks of gestation. She was the second child of phenotypically normal parents. The mother's first pregnancy resulted in the birth of a phenotypically normal boy, and the second and third pregnancies aborted spontaneously. Her birth weight was 2,840 g and her length, 47 cm. Her physical and mental developments were slightly slow. She held her head at three months, sat at nine months, and didn't stand alone at one year of age. She had frequently suffered from infections of the upper respiratory tract.

On physical examinations at the age of one, her height was 75.8 cm, weight 8,690 g, and head circumference 45 cm: all of which were within normal limits.

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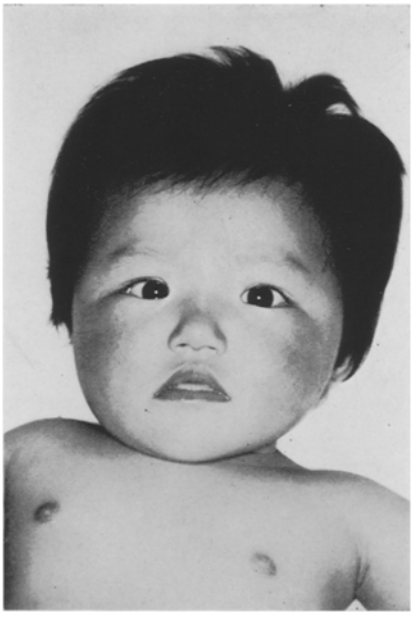

Fig. 1. The appearance of the patient at the age of one.

Her craniofacial stigmata included sloping forehead, slightly large nose, epicanthus, triangular shaped mouth, triangular mandibula, low set ears and flame hemangioma on the forehead (Fig. 1). She had a barrel chest, bilateral simian creases and incurved fifth fingers, and increased muscle tone of the upper extremities. The lower extremities were hypotonic. Heart sounds were clear. She crawled and said meaningless things such as Ma, Mu *etc.* Her development quotient was 87.

Routine laboratory findings were within normal limits. X-ray indicated hypoplasia of the middle phalanx of the fifth fingers, and I.V.P. was normal. EEG was normal, but a moderate hydrocephalus was recognized on the CT scan which showed both dilated side ventricles. ECG was normal. The immunoglobulin levels were normal and the count of T-cells was slightly decreased. Amino acid screening in urine was normal.

DERMATOGLYPHIC STUDY

Dermatoglyphic studies revealed 7 arches and 3 ulner loops on fingers, the total ridge counts was only 19, and b-triradii were absent on both palms. The atd angles were 37.0° on the right palm and 46.0° on the left. There was a simian crease on the left palm. Her mother showed 1 arch, 7 ulner loops and 2 whorles. The total ridge count was 86 and atd angles were 39.0° on the right palm and 41.0° on the left.

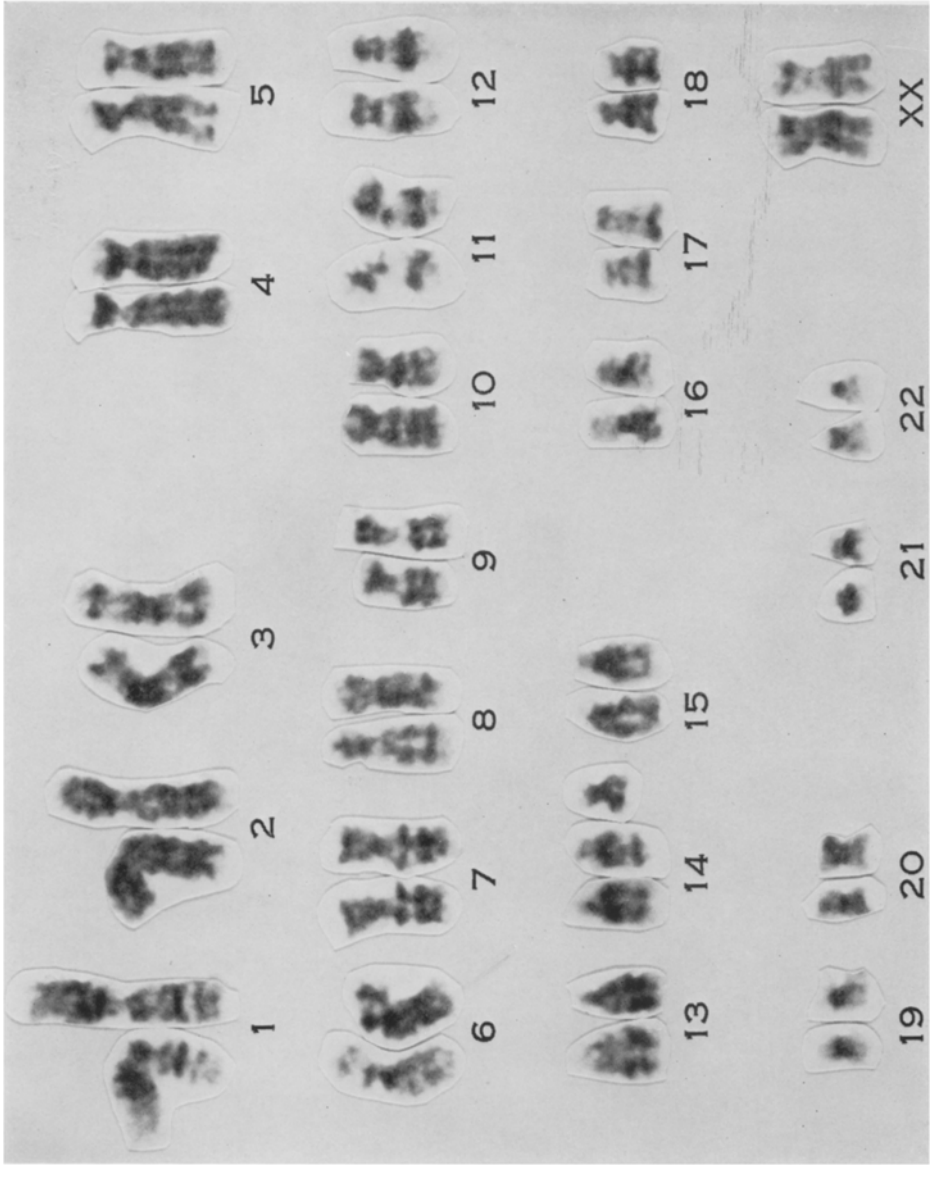

Fig. 2. The karyotype of the patient (G banded).

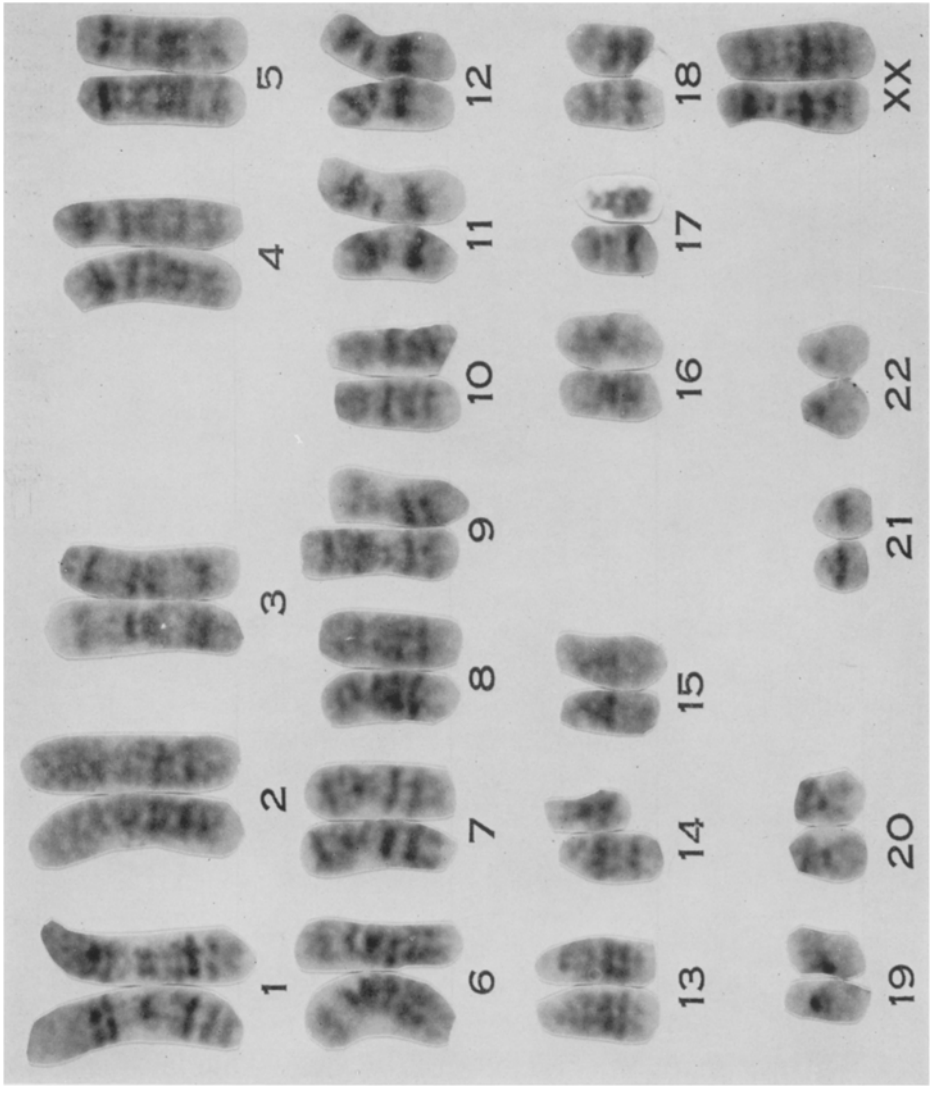

Fig. 3. The karyotype from the mother of the patient (G banded).

CYTOGENETIC STUDY

Chromosome analysis by trypsin Giemsa technique using peripheral blood lymphocytes and cultured skin fibroblasts showed 47 chromosomes with a small extra chromosome. The small extra chromosome was acrocentric and slightly longer than a G chromosome (Fig. 2). Her mother had a reciprocal balanced translocation between No. 9 and No. 14 chromosomes, karyotype being 46,XX, t(9p+;14q−) (Fig. 3). In this patient, the extra chromosome was identified as a der(14) including the short arm, the centromeric region, the proximal part of the long arm of No. 14 and the distal segment of the short arm of No. 9. Her karyotype was 47,XX,+der(14),t(9p+;14q−)mat. The karyotype of the father was normal and her elder brother had 46,XY,t(9p+;14q−) karyotype.

DISCUSSION

Full trisomy 14 seems to be lethal and described in a spontaneous abortion (Kajii *et al.*, 1972). All live-born infants reported to date who had mosaicism (Murken *et al.*, 1970, Rethorè *et al.*, 1975, Martin *et al.*, 1977, Johnson *et al.*, 1979) were severely malformed. Cases with partial trisomy tended to be less affected and seemed to live longer. About twenty cases of partial trisomy 14 have been reported to date outside of Japan, and three cases have been reported other than our case in Japan (Cho *et al.*, 1979, Sakamoto *et al.*, 1980).

The phenotypic symptoms associated with partial 14 trisomy were mental and developmental retardation, recurrent respiratory infection and a complex spectrum of malformation including microcephaly (11/14), broad prominent nose (8/13), wide mouth (9/14), cleft palate (11/14), low set ears (7/14), hypertelorism (4/14) and flexion contracture (6/14) (Johnson *et al.*, 1979). A few cases had cardiac anomaly (Reiss *et al.*, 1972, Short *et al.*, 1972, Muldal *et al.*, 1973, Fryns *et al.*, 1977) and epilepsy (Miller *et al.*, 1979, Short *et al.*, 1972, Muldal *et al.*, 1973, Turleau *et al.*, 1975, Soudek *et al.*, 1978).

In most cases of partial 14 trisomy, one of the parents shows a balanced reciprocal translocation. In the present case, the mother was a balanced translocation carrier between a No. 9 and a No. 14. The patient was a segregant of 3 : 1 disjunction in maternal gametogenesis and had an extra der(14) in addition to a normal female karyotype. The der(14) included the short arm, centromere and proximal part of the long arm of No. 14 chromosome, and the distal segment of the short arm of No. 9 chromosome—the patient was tertiary trisomic for this segment.

Of the 15 cases with partial trisomy 14, 13 cases had translocation carrier parents (4 cases were paternal, 9 cases maternal) (Johnson *et al.*, 1979). In two cases, maternal balanced translocation between No. 9 and No. 14 were reported (Short *et al.*, 1972, Miller *et al.*, 1979). Their symptoms were very similar to those

in our case, but they had microcephalus and seizure which were not observed in our case.

Clinical features seem to relate to the region of the trisomic segment and additional trisomic or monosomic segment of the other chromosomes. Our case was not so severe in mental and developmental retardation and had no major anomaly. It may be because the trisomic segment is a rather small part both of the long arm of chromosome 14 and of the short arm of chromosome 9, though we could not determine where the chromosomal breakpoints were.

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