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PARTIAL DISTAL 12q TRISOMY WITH ARACHNOID CYST

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Summary A three-year-old boy with partial distal 12q trisomy, 46,XY, der(9),t(9;12)(q34.3;q24.31)mat, is described. To our best knowledge, this is the second case in Japan. He has an arachnoid cyst, which has not yet been described in the literature.

INTRODUCTION

There has been 14 reports (16 cases) of partial distal 12q trisomy (Aurias *et al.*, 1978; de Muelenaere *et al.*, 1980; Harrod *et al.*, 1980; Hemming and Brown, 1979; Hirschhorn *et al.*, 1973; Hobolth *et al.*, 1974; Ieshima *et al.*, 1984; Melnyk *et al.*, 1981; Pratt and Bulugahapitiya, 1983; Prieur *et al.*, 1977; Robert *et al.*, 1981; Tajara *et al.*, 1985; Tengström *et al.*, 1985; Zabel and Baumann, 1981). Here we report an additional case of partial distal 12q trisomy which was detected by high resolution chromosome banding technique and review the previously reported patients.

CASE REPORT

The proband was the first product of 28-year-old mother and 30-year-old father, who were unrelated. Because of placental dysfunction, caesarean section was performed at 40 weeks of gestation. Birth weight was 2,055 g, the length 44.2 cm, and the head circumference 31.0 cm. The early neonatal period was complicated with hyperbilirubinemia, hypoglycemia and feeding difficulties. He had also retinopathy of prematurity. He controlled his head at 6 months of age, rolled over at 9 months, and sat alone at 1 year 2 months. At the age of 3 years 1 month, his body weight was 10.9 kg (−1.8 SD), the length 90.3 cm (−0.7 SD), and the head circumference 44.4 cm (−3.5 SD). His muscle tone was decreased. Main clinical features were severe mental retardation (DQ 23), microcephaly, brachycephaly, flat facial profile,

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hypertelorism, antimongoloid slant, blepharoptosis, broad eyebrows, flat nasal bridge, anteverted nares, long philtrum, carp mouth, pointed chin, short tapering fingers, and overlapping toes (Fig. 1).

Dermatoglyphics was abnormal; low TFRC(47) and bilateral absent digital triradius c. Sydney line was noted on the left hand (Table 1). Skeletal survey revealed normal. IVP was normal. CT scan of the brain showed an arachnoid cyst at the left middle cranial fossa, mild dilatation of the lateral ventricles, and asymmetrical Sylvian fissures (Fig. 2). EEG was normal.

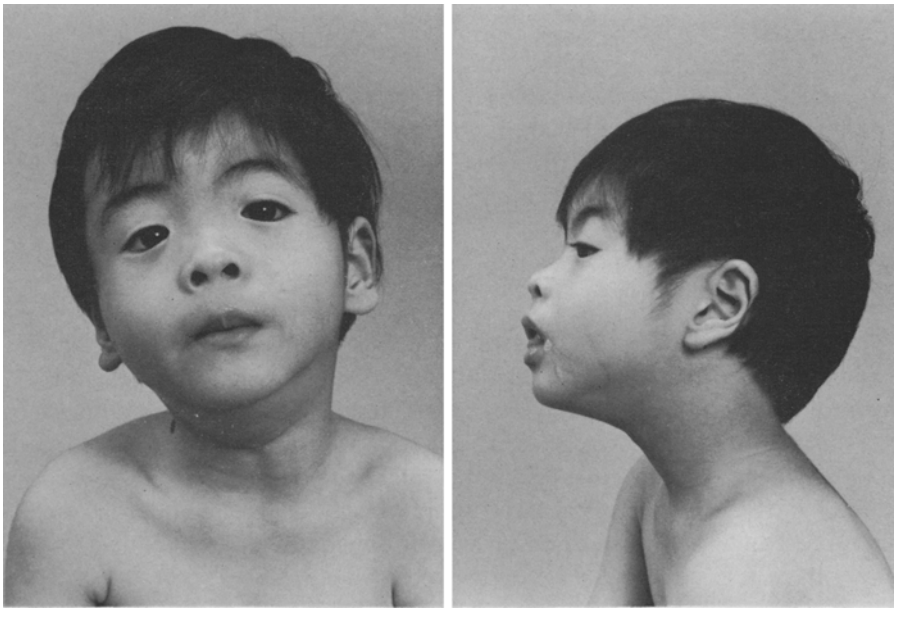

Fig. 1. The patient at 3 years of age.

Table 1. Dermatoglyphic findings of the proband.

	1	2	3	4	5
Left hand	U	T	U	U	U
Right hand	U	U	U	U	U
Palmar formula					
Left: 11.0.7.3.13′	-t′	-0.0./0.0.0.0.			
Right: 11.0.7.3.13′	-t′	-0.0./0.0.0.0.			

U, ulnar loop; T, tented arch.

CYTOGENETIC FINDINGS

The karyotype of the proband was 46,XY,der(9),t(9;12) (q34.3;q24.31)mat. The mother's karyotype was 46,XX,t(9;12) (q34.3;q24.31). The father showed normal male karyotype (Fig. 3).

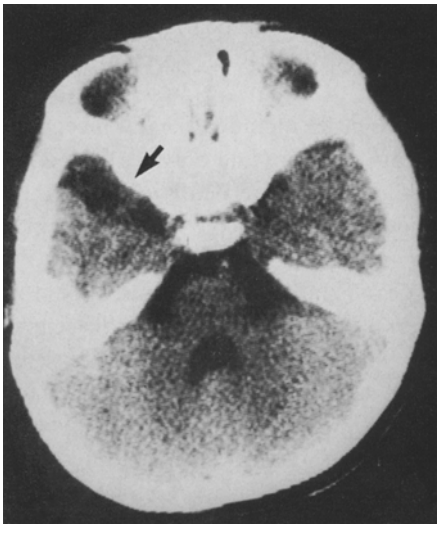

Fig. 2. CT scan of the brain at 3 years of age. The arrow indicates the arachnoid cyst.

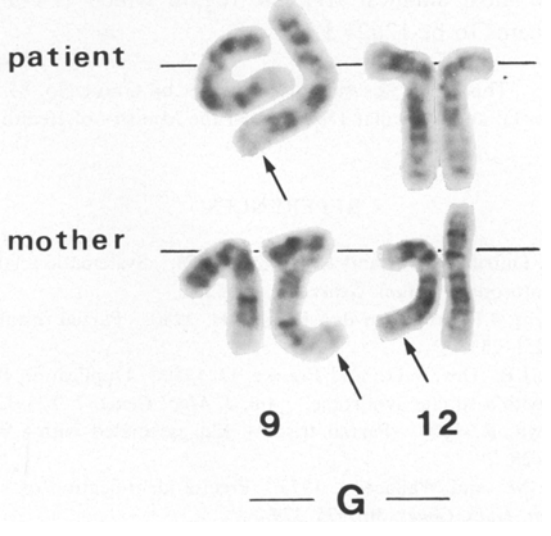

Fig. 3. Partial karyotype of the proband and his mother with high resolution G banding. Breakpoints are at 9q34.3 and at 12q24.31.

DISCUSSION

Partial distal 12q trisomy is characterized by mental retardation, growth retardation, dyscephaly (dolichocephaly or brachycephaly), hypertelorism, flat nasal bridge, low-set ears, poor lobulation, down-turned mouth, micrognathia or pointed chin, short neck, loose skin at nape, wide set nipples, simian creases, genitourinary anomalies, sacrococcygeal dimple, and heart defect. Although the present case showed almost similar craniofacial features, visceral anomalies and sacrococcygeal anomalies were not observed.

CT findings in 5 previous cases included frontal lucent areas, mild hydrocephaly, cortical atrophy, dilated third and/or lateral ventricles and cisterna magna. Two autopsy clarified dysplasia of cerebellar dentate nucleus, hydrocephaly, and brain malformation. An arachnoid cyst, noticed in the present case, has not been described so far.

Fourteen out of 17 cases, including ours, resulted from malsegregation of parental translocation (10 maternal, 4 paternal). The 12q segment was translocated to a different chromosome; 9(five cases), 4(three cases), 2(two cases), 17(two cases), 18(one case), 21(one case). The remaining two cases had an intrachromosomal duplication (Harrod *et al.*, 1980). One case had an inverted duplication (Ieshima *et al.*, 1984). Trisomic segments described in the present case and the previous reports are variable; 12q24 → qter(four cases), 12q24.1 → qter(two cases), 12q24.13 → qter(one case), 12q24.2 → qter(two cases), 12q24.3 → qter(two cases), 12q24.31 → qter(two cases), 12q24.1 → q24.33(two cases), 12q12 → qter(one case), 12q21.2 → qter(one case). The most smallest trisomic region which is compatible with 12q trisomy syndrome seems to be 12q24.3.

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