A FEMALE INFANT WITH PURE DUPLICATION 12q24.2 → qter

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Summary A female infant with pure partial trisomy 12q, 46,XX, inv dup(12)(q24.2 → qter), is described. This is the first report from among oriental people. Several skeletal anomalies hitherto not described and dermatoglyphics are presented in detail.

INTRODUCTION

12q Trisomy is a rare condition and only ten cases have been reported so far (Hobolth *et al.*, 1974; Hemming and Brown, 1979; Harrod *et al.*, 1980; de Muelenaere *et al.*, 1980; Roberts *et al.*, 1981; Zabel and Baumann, 1981; Melnyk *et al.*, 1981; Pratt and Bulughapitiya, 1983). The remarkable similarity in clinical features between them suggests that trisomy 12q is a clinically recognizable syndrome. Seven of these have arisen as a result of a familial translocation and two as a result of duplication of distal region of 12q in mosaicism. Here we report a female infant with pure distal trisomy 12q.

CASE REPORT

The proband was the second child of healthy and unrelated 27-year-old mother and 27-year-old father. She was born at 41 weeks of gestation after normal pregnancy and delivery except for poor fetal movement. Birth weight was 3,083 g, the length 51 cm, and the head circumference 32.5 cm. The face was dysmorphic: brachycephaly, left exotropia, hypertelorism, long eye lashes, flat nasal bridge, small nose with hypoplastic nasal tip, downward corners of the mouth, mild micrognathia

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and poorly lobulated ears. Short neck and widely spaced nipples were also noted. The hands were small with mild brachydactyly and hypoplastic nails. The thumbs and the first toes were big and broad (Fig. 1). Small ventricular septal defect was suspected by electrocardiogram and echocardiogram.

Dermatoglyphics showed ridge dissociation in bilateral fifth fingers, decreased a-b ridge count (63), and third interdigital patterns (Table 1). Dermatoglyphics of the parents could not be studied.

Broad distal phalanges of thumbs and first toes, and disproportionally advanced

Fig. 1. Proposita at the age of 1 year.

Table 1. Dermatoglyphic findings of proposita. Finger pattern and ridge count.

	1	2	3	4	5
Left hand	W	R	A	U	D
ridge count	22	4	0	11	...
Right hand	W	R	U	U	D
ridge count	26	5	5	14	...
Palm formula					
Left: 9.9.5".4.13'. -t-0.0./0.0.L.0.					
Right: 9.9.5'.3.13'. -t-0.0./0.0.L.0.					

W, whorl; R, radial loop; U, ulnar loop; A, arch; D, dissociated ridge.

Fig. 2. Radiogram of the hands and feet at 1 year of age. Note the broad distal phalanges of the thumbs and the great toes, and advanced bone age in the tarsal and carpal bones.

bone age in carpal and tarsal bones were noticed by radiological study (Fig. 2). CT scan of the brain at three months showed mild dilation of third and lateral ventricles, and cortical atrophy.

She suffered from frequent respiratory infections. Her physical and psychomotor development was retarded. At the age of 1 year, her body weight was 5,960 g (-3.1 SD), the length 65.9 cm (-3.0 SD) and the head circumference 44.0 cm (-1.0 SD). She controlled her head at six months of age, rolled over at one year, but could not sit alone yet. Developmental quotient was 30 at one year.

CYTOGENETIC FINDINGS

The karyotype of the proband was determined to be 46,XX,inv dup(12)(q24.2→qter) by R- and G-banding studies (Fig. 3). The karyotypes of the parents were normal by R-banding method.

Fig. 3. Partial karyotypes of chromosomes 12 after high resolution R-banding (Left) and G-banding (right).

Table 2. Clinical and cytogenetic findings of partial trisomy 12q.

	1. (1974)	2. (1979)	3. (1980)	4. (1980)	5. (1981)	6. (1981)	7. (1981)	8. (1983)	9. (1984)	
Age at the report	2 Y	6 days	3 Y	1 Y	20 Y	10 M	2.5 M	1Y 4 M	1 Y	
Sex	male	female	female	male	female	male	male	male	female	
Karyotype	der(21), t(12;21) (q24;p11)	der(18) t(12;18) (q24.1;q24.1; qter)	dup(12) (q24.1→q24.5) mosaic	der(17) t(12;17) (q24;q25)	der(17) t(12;17) (q24;q25)	der(2) t(2;12) (q37; q24.1)	der(9) t(9;12) (q24.3; q24.1)	der(4) t(4;12) (p16; q24.1)	der(9) t(9;12) (p24; q21.1)	dup(12) (q24.2→ qter)
Psychomotor retardation	+	...	+	+	+	+	+	+	+	10/10
Growth retardation	+	...	+	+	+	+	+	+	+	10/10
Cardiac defect	+	-	-	-	-	-	+	+	+	7/11
Hypertelorism	+	+	+	+	-	-	+	+	+	8/11
Dyscephaly	B	B	D	D	-	D	B	Asym	D	9/11
Flat nasal bridge	-	+	+	+	-	+	+	+	+	8/11
Down-turned mouth	-	+	+	+	+	-	+	+	-	8/11
Micrognathia	+	-	+	+	-	+	+	+	+	8/11
Low-set ears	+	-	+	+	-	+	+	+	+	7/11
Poorly lobulated ears	+	+	+	+	-	+	+	-	+	8/11
Short neck	-	+	+	+	-	+	+	+	+	8/11
Loose skin of the nape	+	+	+	+	-	+	+	+	+	8/11
Wide-set nipple	-	+	+	+	-	-	+	-	+	7/11
Sacral dimple	-	+	-	+	-	+	+	+	+	7/11
Cryptorchidism	+	...	...	+	...	+	+	+	...	5/5
Simian crease	+	+	+	+	-	+	+	+	-	7/11

B, brachycephaly; D, dolichocephaly; Asym, facial asymmetry. 1. Hobolth *et al.* (1974); 2. Hemming and Brown (1979); 3. Harrod *et al.* (1980); 4. de Muelenaere *et al.* (1980); 5. Roberts *et al.* (1981); 6. Zabel and Baumann (1981); 7. Melnyk *et al.* (1981); 8. Pratt and Buluga-hapitiya (1983); 9. present case.

DISCUSSION

The clinical features and cytogenetic findings of 11 cases with 12q trisomy were reviewed (Table 2). Trisomy 12q is characterized by psychomotor retardation, growth retardation, dyscephaly, hypertelorism, flat nasal bridge, down-turned mouth, micrognathia, low-set ears, poor lobulation of ears, short neck, loose skin of nape, wide-set nipples, sacral dimple, cryptorchidism and simian crease. Hydrocephaly or ventricular dilation was occasionally observed. Normal or big head associated with sacral dimple may have a diagnostic value in trisomy 12q, and these features may indicate the neural tube defect and abnormal liquor kinetics. The present case was found to have many clinical features similar to those described in the Caucasian cases. These characteristic features are well observed in infants or young cases, but gradually become obscure with age in adult cases.

Short limbs (three cases), planovalgus feet (four cases), ulnar deviation of hands (four cases), brachymetacarpus (two cases) and camptodactyly (one case) were observed in the reported cases. Present case showed small hand with brachydactyly, big thumbs and toes, and disproportionally advanced bone age. Skeletal abnormalities of extremities may be a characteristic feature in trisomy 12q syndrome.

Break point was distributed in the region of 12q(12q21.1→q24.3). Present case was inv duplication 12q24.2-qter, therefore this is the first case of pure trisomy 12q.

Ridge dissociation or ridge hypoplasia has been reported in two cases with 12q trisomy (Melnyk *et al.*, 1981; Roberts *et al.*, 1981). The finger tip patterns are not consistent; increased arches or increased whorls. The a-b ridge count or total ridge count was not mentioned. In our case, ridge dissociation and decreased a-b ridge count was observed. Poor ridge formation may be a characteristic in trisomy 12q.

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