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A CASE OF AN INTERSTITIAL DELETION OF THE LONG ARM OF CHROMOSOME 5: 46,XX,del(5)(q15q22)

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Summary A female infant with mild mental retardation and dysmorphic features was found to have an interstitial deletion of the long arm of chromosome 5: 46,XX,del(5)(q15q22).

INTRODUCTION

To our knowledge, 7 cases with an interstitial deletion of the long arm of chromosome 5 have been reported. Here is an additional case and the karyotype-phenotype relationship is discussed.

CASE REPORT

The proposita was born after an uneventful course of pregnancy and normal delivery. Her birth weight was 2,840 g, length 48 cm, and head circumference 32.5 cm. Her mother and father, who were healthy and nonconsanguineous, were 30 and 29 years of age at her birth. There was no family history of mental retardation or congenital abnormalities. At birth, bilateral club feet, which were corrected by 6 months of age, were noticed. Her motor development was normal: head control at 4 months, sitting at 6 months, and walking alone at 12 months. However, she could speak only about 10 words until 2 years of age.

When she was referred to our hospital at 26 months of age, her weight was 10.4 kg (−0.9 SD), length 81.1 cm (−1.6 SD), and head circumference 46.3 cm (−0.9 SD). Her clinical features included narrow forehead, frontal bossing, epicanthus, blepharophimosis, puffy eyelids, anteverted nostrils, prominent nasal alae, downturned mouth with thick lips, retromicrognathia, short neck, wide chest, and clinodactyly of 5th fingers (Fig. 1).

Dermatoglyphics was abnormal: 7 whorls on finger tips, simian crease (bil), hypothenar pattern (1t), termination of mainline A to area 1 (rt), and intermediate axial triradius (t') (1t) (Fig. 2). Intravenous pyelography showed the horse-shoe kidney.

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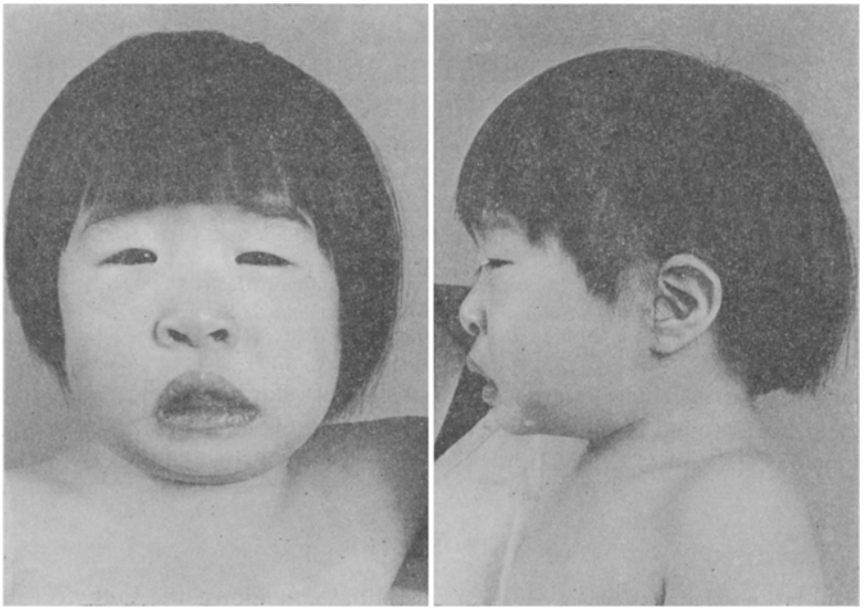

Fig. 1. Front and side view of the present case at 26 months of age.

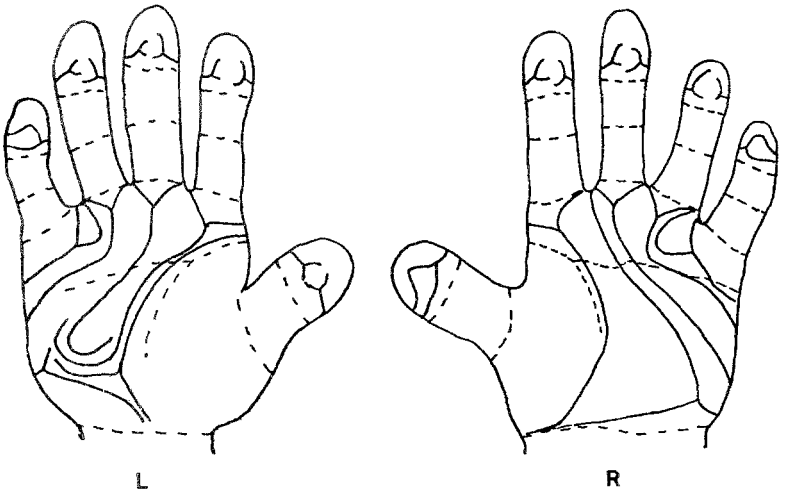

Fig. 2. Dermatoglyphics of the present case.

Her developmental quotient was 89 at 26 months of age.

Chromosome study with high resolution G-banding method showed an interstitial deletion of the long arm of chromosome 5; 46,XX,del(5) (q15q22) (Fig. 3). Both parents had normal karyotypes.

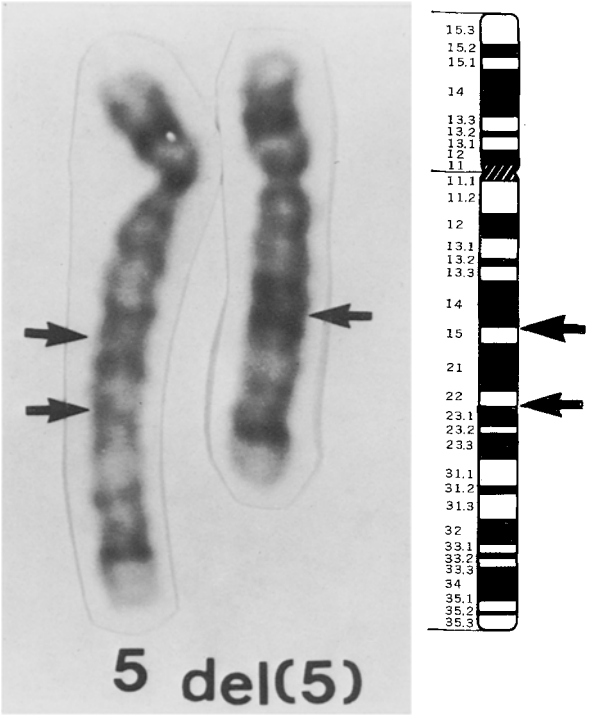

Fig. 3. High-resolution G-banded chromosome 5 from the present case. The left homolog is normal but the right one is affected with a deletion in 5q15 through 5q22. Arrows show the break-points.

DISCUSSION

Only 7 cases with an interstitial deletion of the long arm of chromosome 5 have been reported so far. Although there have been no distinct clinical features of 5q− cases, the abnormalities such as mental retardation, short stature, narrow forehead, frontal bossing, epicanthus, flat nasal bridge, anteverted nostrils, retromicrognathia, malformed ears, club foot, renal anomaly and abnormal dermatoglyphics (simian crease, increased whorls, and hypothenar pattern) have been frequently observed (Table 1).

Figure 4 showed the deleted segments in 5q− cases. No direct relationship between the severity and the variation of clinical features and the amount and/or the location of deleted segments was observed.

Table 1. Clinical findings of patients with 5q−.

Case	(1)	(2)	(3)	(4)	(5)	(6)	(7)	(8)	Total
Sex	F	F	F	F	M	M	F	F	
Birth weight (g)	2,900	4,050	3,460	2,500	3,300	2,980	3,170	2,840	
Age mother/father	28/39	36/35	29/31	21/25	32/33	25/25	27/37	30/29	
Mental retardation	+	+	+	+	+	+	+	+	8/8
Muscle tone		hyper			hypo		hypo	normal	
Short stature	+	+	+	+	+	+	−	+	7/8
Microcephaly	−	−	+	+	−	+	+	−	4/8
Brachycephaly		+		−		+	+	−	3/5
Narrow forehead		+		+	−*	+	−	+	4/6
Frontal bossing	+	−	+*	+	+		+	+	6/7
Hypertelorism		−*	+	+	−*	+	+	−	4/7
Epicanthus	+	+			+		+	+	5/5
Flat nasal bridge	+	−	+	+	+	−	+	+	6/8
Anteverted nostrils	+	−		+	−	+	−	+	4/7
Cleft palate	+	−	+		−	+		−	3/6
Large philtrum				+		+	+	−	3/4
Retromicrognathia			+	+		+	−	+	4/5
Malformed ears	+	−	+	+	+	+	+	−	6/8
Short neck	+*		+	+		+*	+	+	6/6
Clinodactyly				+	−			+	2/3
Club foot	+	+			−	+		+	4/5
Renal anomaly	+	−	+		−			+	3/5
Excess of whorl pattern	+			+		+	−	+	4/5
Hypothenar pattern	+	−		−	−	+	+	+	4/7
Simian crease	+	−		−	−	+	+	+	4/7

* Judged from photographs. (1) Centerwall *et al.* (1978). (2) Pescia *et al.* (1978). (3) Felding *et al.* (1980). (4) Stoll *et al.* (1980). (5) Silengo *et al.* (1981). (6) Ohdo *et al.* (1982). (7) Rodewald *et al.* (1982). (8) Present case.

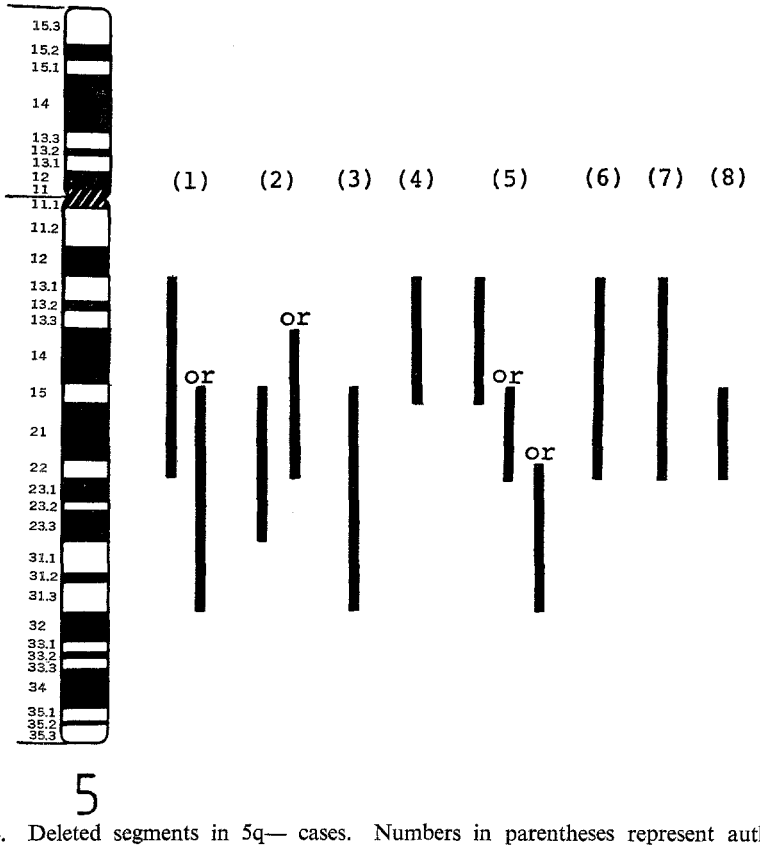

Fig. 4. Deleted segments in 5q— cases. Numbers in parentheses represent authors shown by the same number in Table 1.

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