Sibs lacking characteristic features of duplication of distal 17q

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SUMMARY Two brothers with karyotype 46,XY,-16,+der(16),t(16;17)(q24.3;q25.1)pat are presented. It is commonly thought that duplication of distal 17q results in a clinically recognisable syndrome. Although our cases had several features often seen in patients with autosomal chromosome aberrations, they did not have any of the specific features found in other patients with this duplication.

It is known that there are phenotypic similarities in all cases of duplication of distal 17q reported so far.¹⁻¹¹ Our cases, however, had no specific features characteristic of duplication of distal 17q. Possible reasons for this are discussed.

Case reports

CASE 1

The proband, a male infant, was born at term in March 1988 and was the second child of healthy and

unrelated parents; the father was 38 and the mother 36 years old. The older brother (case 2) of the proband is mentally retarded. The mother has had no spontaneous abortions.

His birth weight was 3170 g. He was referred to our outpatient clinic in April 1988 because of poor weight gain. At that time height was 50.0 cm (-1.8 SD), weight 3480 g (-2.3 SD), and head circumference 36.0 cm (-1.0 SD). Physical findings included: hypertelorism, poorly delineated and long philtrum, bilateral preauricular pits, right testis in the inguinal canal, and bilateral palmar transverse creases (fig 1). The thorax, including heart and lungs, and the abdomen were clinically normal. The results of routine biochemical and haematological examinations were all within the normal range. Echocardiography showed no congenital heart defects and CT scan of the brain showed no evidence of structural abnormalities. His chromosomes and those of his parents were analysed from cultured lymphocytes. The father's karyotype was 46,XY,t(16;17)(q24.3;q25.1) and that of the proband

FIG 1 Facial appearance of proband at two months.

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FIG 2 Partial karyotypes of father, proband (case 1), and brother (case 2). Arrows indicate breakpoints on the normal 16q and 17q. An extra dark band (17q25.1→qter) was noted in the terminal region of 16q on the right.

FIG 3 Facial appearance of proband's brother (case 2) at six years eight months.

was 46,XY,-16,+der(16),t(16;17)(q24.3;q25.1)pat (fig 2). The mother's karyotype was normal.

At six months DQ was about 50.

CASE 2

The older brother of the proband was born at term in August 1981. Birth weight was 2800 g. Since March 1987, he has been receiving treatment for epilepsy. He has been attending a special school for the mentally retarded since April 1987.

When he visited our outpatient clinic in May 1988, his height was 98.3 cm (-4.7 SD), weight 18.1 kg (-1.9 SD), and head circumference 48.9 cm (-2.2 SD). Physical examination showed the following abnormalities: esotropia of the right eye, hypertelorism, poorly delineated and long philtrum, low set ears, and bilateral talipes valgus (fig 3). The lungs, heart, and abdomen were normal. *Atd* angle on both palms was 50°. Laboratory tests showed normal values. Echocardiography yielded normal results. CT scan of the head showed dilatation of the lateral ventricles. His IQ was about 25. His karyotype was 46,XY,-16,+der(16),t(16;17)(q24.3;q25.1)pat (fig 2).

Discussion

Among all the cases with duplication of distal 17q reported so far,¹⁻¹¹ two cases were presumed to have monosomy extensive enough to affect the clinical picture,^{2,5} while all other cases, including ours, were not. In spite of the differences in the size of the chromosome segments involved, there are phenotypic similarities in all the cases with duplication of distal 17q. Among these features, those which are seldom noted in cases with other autosomal chromosome aberrations are as follows: widow's peak, temporal retraction, neck webbing, low posterior hairline, proximal limb shortness, and polydactyly or syndactyly or both. In cases with duplication of the same chromosome segments as in our cases, several of these characteristic features were seen.^{8,9} However, our cases have none of these characteristic features (table). We have no satisfactory explanation for this discrepancy. It may be that in our cases the size of the duplication of distal 17q is smaller than in the other cases.^{8,9} Another possibility might be that chromosome studies are only reported in cases with many minor

TABLE The extent of duplication of distal 17q and associated clinical features in previously reported cases and present cases. (Revised from Naccache et al.⁶)

	References											Present cases	
	1	2	3	4	5	6	7	8	9	10	11	Proband	Brother
Extent of duplication	q23→qter	q23.1→qter	q22→qter	q21→qter	q23→qter	q23→qter	q12→qter	q25→qter	q25.1→qter	q23.1→qter	q21.1→qter	q25.1→qter	
Sex	F M	F M	M	F	M	M	M	M	M	F	F	M	M
Short stature	+	+	+	+	+	+	+	+	+	+	+	+	+
Psychomotor retardation	+	+	+	+	+	+	+	+	+	+	+	+	+
Microcephaly	+	+	+	+	+	+	+	+	+	+	+	+	+
Frontal bossing	+	+	+	+	+	+	+	+	+	+	+	+	+
Temporal retraction	+	+	+	+	+	+	+	+	+	+	+	+	+
Facial asymmetry	+	+	+	+	+	+	+	+	+	+	+	+	+
Widow's peak	+	+	+	+	+	+	+	+	+	+	+	+	+
Hypertelorism	+	+	+	+	+	+	+	+	+	+	+	+	+
Antimongoloid slant	-	-	+	-	+	+	+	+	+	+	+	+	+
Epicanthus	+	+	+	-	+	+	+	+	+	+	+	+	+
Flat nasal bridge	+	+	+	-	+	+	+	+	+	+	+	+	+
Wide mouth with thin upper lip	+	+	+	+	+	+	+	+	+	+	+	+	+
Downturned corners of the mouth	+	+	+	+	+	+	+	+	+	+	+	+	+
Cleft lip and/or palate	+	+	+	+	+	+	+	+	+	+	+	+	+
Highly arched palate	-	-	-	-	+	+	+	+	+	+	+	+	+
Micrognathia	-	-	+	+	+	+	+	+	+	+	+	+	+
Low set, posteriorly rotated ears	+	+	+	+	+	+	+	+	+	+	+	+	+
Short neck	-	-	+	+	+	+	+	+	+	+	+	+	+
Webbed neck	+	+	+	+	+	+	+	+	+	+	+	+	+
Low posterior hairline	+	+	+	+	+	+	+	+	+	+	+	+	+
Widely spaced nipples	-	+	+	+	+	+	+	+	+	+	+	+	+
Undescended testis	-	+	+	-	+	+	+	+	+	+	+	+	+
Proximal limb shortness	-	+	+	-	+	+	+	+	+	+	+	+	+
Polydactyly of hands/feet	-	-	+	-	+	+	+	+	+	+	+	+	+
Syndactyly of fingers/toes	+	-	+	+	-	+	+	+	+	+	+	+	+
Hyperlaxity of limb joints	+	+	+	+	+	+	+	+	+	+	+	+	+
Heart defects	+	+	+	+	+	+	+	+	+	+	+	+	+
Kidney anomalies	-	+	+	+	+	-	-	+	+	+	+	+	+
CNS abnormalities	+	+	+	+	+	+	+	+	+	+	+	+	+

*Not described or unclear. HM=heart murmur. NE=not examined.

physical abnormalities, and not in cases with few of these features, such as ours. Furthermore, cases with de novo tandem duplication of distal 17q who have no specific features have not been published, since a confident diagnosis cannot be made at present. Therefore, a considerable number of patients may exist with duplication of distal 17q who have no constellation of specific features noted in previously reported cases.

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