Case reports

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A child with partial trisomy of chromosome 17 and partial monosomy of chromosome 3: 46,XY,der(3),t(3;17)(p25;q23)

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SUMMARY A male infant with multiple congenital abnormalities and global retardation was found to have a translocation resulting in partial trisomy for the distal end of the long arm of chromosome 3. The phenotypically normal father carried a balanced reciprocal translocation between chromosomes 3 and 17.

A number of cases have been described with partial trisomy 17q, but none is associated with partial monosomy 3p. The child presented below has clinical features similar to those found in partial trisomy for 17q, whereas the contribution of the small 3p deletion is less easy to ascertain.

Case report

The proband, a male infant, was born after a normal

pregnancy, by Caesarian section for breech presentation, to a 20-year-old primigravid mother and 24-year-old father. The parents were non-consanguineous, healthy, and of normal intelligence. Physical examination (fig 1) revealed a large anterior fontanelle, wide sagittal suture, narrow biparietal diameter, low set malrotated ears, mongoloid slant to the eyes, epicanthic folds, hypertelorism, broad nasal bridge, high arched palate, wide mouth, micrognathia, low posterior hairline, webbed neck, short proximal limbs, postaxial polydactyly of hands and feet, sacral dimple, cryptorchidism, rocker bottom feet, and a heart murmur. An intravenous pyelogram, electrocardiogram, and ultrasound of the ventricles were normal. Skeletal survey revealed abnormal sclerosis of the long bones, rudimentary bilateral sixth fingers, poor mineralisation of the skull vault, and a wide 'short chest'.

Review at 19 months revealed head circumference 46.2 cm, height 70 cm, and weight 8.11 kg (<3rd

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FIG 1 The proband.

centile), with little change at 30 months. Developmental assessment on the latter occasion showed significant delay, with all skills at a 5-month level.

CYTOGENETIC FINDINGS

Peripheral blood lymphocytes were cultured from the proband and both parents for chromosome studies with trypsin G banding. The proband showed an unbalanced chromosome rearrangement involving chromosome 3 (fig 2). Examination of the paternal chromosomes showed a balanced translocation between the short arms of chromosome 3 and the long arms of chromosome 17 (fig 2). His karyotype was 46,XY,t(3;17)(p25;q23) and that of the proband 46,XY,der(3),t(3;17)(p25;q23), resulting in partial monosomy of 3p and partial trisomy of 17q. The maternal chromosomes were normal.

Discussion

This child has partial trisomy for 17q22→17qter and partial monosomy for 3p25→3pter, a combination not previously described.

Partial trisomy for 17q is rare. A patient with pure trisomy for this section of 17q has been described by Fryns *et al*,¹ while the first report was that of Fouquette *et al*² who, in 1974, identified a duplication of 17q using fluorescence. Berberich *et al*³ reported three affected persons in a family where a 7q;21q reciprocal translocation was segregating.

FIG 2 Partial karyotypes of father and proband. (a) Paternal balanced translocation (3;17)(p25;q23). (b) Proband's unbalanced translocation der(3),t(3;17)(p25;q23) pat.

TABLE Comparison of clinical features in present case with those previously reported.

	Present case	Cases of trisomy 17q22→17qter ^{1 3 4}	Other cases of 17q duplication ^{2 5}
Mental retardation	+	5/5	2/2
Growth retardation	+	5/5	1/2
Long head, narrow anteriorly	+	4/5	1/2
Wide open sagittal suture	+	—	—
Low set malrotated ears	+	4/5	2/2
Epicanthic folds	+	3/5	1/2
'Squinty' eyes	—	4/5	1/2
Wide mouth	+	4/5	2/2
High arched or cleft palate	+	4/5	2/2
Micrognathia	+	2/5	1/2
Webbed neck	+	4/5	1/2
Low posterior hairline	+	5/5	0/2
Polydactyly	+	1/5	1/2
Short proximal limbs	+	1/5	0/2
Cryptorchidism	+	2/3	0/1
Congenital heart disease	—	2/5	2/2
Renal anomaly	—	2/5	0/2

Turleau *et al*⁴ described a child with trisomy for the same region of 17q and a deletion of 5p. The main clinical features in these subjects are summarised in the table. A number of features are common to all. Also included in the table is the child described by Fouquette *et al*² and one reported by Salamance-Gomez and Armendares.⁵

The only description of a subject monosomic for the distal part of 3p is an infant described by Verjaal and De Nef.⁶ The clinical features of this child were quite distinct from those of our case, apart from the abnormal ears, high palate, and undescended testes, common to many chromosomally abnormal infants.

This case supports the suggestion of Turleau *et al*⁴ that trisomy 17q22→17qter is a recognisable condition whose chief features are a narrow head, wide mouth, neck webbing, low posterior hairline, and polydactyly.

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the short arm of chromosome 3. *Am J Dis Child* 1978; **132**:43-5.

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De novo translocation heterozygote with three reciprocal translocations

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SUMMARY An extremely rare case of a child with three balanced reciprocal translocations involving six different autosomes is described. These abnormalities have apparently arisen de novo and seem to have only relatively minor phenotypic effects. The meiotic possibilities are discussed and cytogenetic markers suggest that the damage may have occurred in a paternal gamete.

It is rare to find more than a single translocation in a person¹ and extremely rare to find more than two.² Complex chromosome rearrangements involving exchanges between three or more chromosomes in a circular fashion show certain similarities to the present case, in that as many as four or five derived chromosomes can be seen in the karyotype of the carrier.³⁻⁴ The observation of six de novo abnormal chromosomes in a live-born child is unrecorded to the best of our knowledge.

Case report

This boy presented at the age of 9 years because of maternal anxiety about his short stature. It was reported that he had always been short and had been overtaken by his sister who is 2 years younger at the age of 5 years. At the age of 10 years his height was 4 cm below the 3rd centile, and his height velocity over the previous year had been just below the 10th centile. Radiology at this time showed his bone age to be about 2 years delayed.

Despite this problem he was found to be a healthy, well proportioned boy whose performance at school

was average. He showed no indications of malabsorption. Both his parents and his two sisters were of average height and no one else in the family was reported to have short stature. There was no evidence of consanguinity.

The mother appeared to have had a normal uneventful pregnancy and an induced birth at 38 weeks' with normal delivery. At birth the child was found, at 2497 g, to be 100 g below the 10th centile. The neonatal period was normal and developmental progress was satisfactory, if a little slow. His intelligence remained normal but his speech was rather slow and he had been given speech therapy at 4 years of age.

Materials and methods

Chromosome preparations were made from conventional phytohaemagglutinin stimulated cultures of whole blood. The metaphases were G banded⁵ and sequentially C banded.⁶

Results

One hundred cells were examined revealing a grossly abnormal karyotype: 46,XY,t(1;7)(q42;q22), t(5;9)(q31;q32),t(13;16)(q21;q22) (figs 1 and 2). The three translocations appeared to be reciprocal and balanced. Parental karyotypes were normal.

Comparison of C band polymorphisms showed that the derived chromosomes der (1), der (9), and der (16) were of paternal origin while the normal homologues were from the mother (fig 2). In addition, the mother had a marker chromosome 13ps+ which was unaltered in the child, while the father had a marker chromosome 5ph+ which could be identified as the derived chromosome 5 in the child