Three children with partial trisomy 1q and partial monosomy 3p

G T McCARTHY*, C N FEAR†, AND A C BERRY†

From *the Royal Alexandra Hospital for Sick Children, Dyke Road, Brighton, Sussex BN1 3JN; and †the SE Thames Regional Genetics Centre, Guy's Hospital, St Thomas Street, London SE1 9RT.

SUMMARY We report a kindred in which three children suffer from partial trisomy 1q and partial monosomy 3p, transmitted by a balanced translocation which is maternal in one family and paternal in the other. The clinical features of the three children are similar and include severe mental handicap and severe scoliosis in the older two.

Case reports

The family pedigree is shown in fig 1. Case 1 (III.4) was a girl, born in 1965. Case 2 (III.5) was a boy, born in 1966. Case 3 (III.2) was a girl, born in 1967. The clinical features of the three are shown in the table and illustrated in fig 2. The beaked nose and broad thumbs initially suggested a diagnosis of Rubinstein-Taybi syndrome.

CYTOGENETIC STUDIES

Cases 1 and 2 were karyotyped in 1968 and Case 3 in 1970, all using orcein stain. No abnormality was detected. In 1976, the children were reinvestigated using standard Giemsa banding. They were each found to have unbalanced chromosome complements giving rise to partial trisomy 1q and monosomy for the terminal part of 3p. The parents were investigated and the mother of cases 1 and 2 and the father of case 3 (who were themselves brother and

sister) carried a balanced reciprocal translocation: 46,XX or XY,t(1;3)(q25;p23) (fig 3). The other parent in each family was chromosomally normal. Cases 1, 2, and 3 have the chromosome complement: 46,XX or XY,der(3)(1;3)(q25;p23).

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FIG 1 Pedigree showing segregation of translocation.

FIG 2 Cases 1 (left) and 2 (right) aged 20 and 19 years respectively.

TABLE Clinical features of three cases.

	Case 1	Case 2	Case 3
Birth weight (kg)	2.73	3.01	2.84
Pregnancy	Normal	Toxaemia	Hypertension
Intercurrent illness	Pyloric stenosis, Ramstedt's operation	—	Bloody diarrhoea. Rx gluten free diet
Mental retardation	Severe. Self care but no speech	As case 1	Severe
Face			
Beaked nose	+	+	+
Low set ears	+	+	—
Brushfield spots	—	—	+
Low posterior hair line	+	+	+
Broad thumbs	+	+	+
Big toe	Broad	Broad	Short
Testes	—	Undescended	—
Adult height (cm)	135	135	NK
Adolescent kyphoscoliosis	++	++	NK

NK=not known.

FIG 3 Partial karyotype showing balanced translocation.

Discussion

The family is reported since they appear to be chromosomally unique, and the development of scoliosis in the two sibs is striking. There are two known cases with a similar chromosome abnormality, but with different though neighbouring breakpoints. In one,¹ the unbalanced translocation appears to have arisen de novo, whereas in the other² the mother carried the reciprocal translocation. The case described by Yunis *et al*¹ shows little in common with our cases but was reported during infancy. The case of Schinzel² was reported as an adult and showed severe mental handicap and a mild kyphoscoliosis, but otherwise had few similarities

with our cases. He had striking hypertrichosis which is not a feature of the children described here. These three children appear to be the only ones described with their particular chromosome abnormality. The family have previously contributed to a study localising the gene for peptidase C to the distal end of 1q.³ Our report indicates how readily cytogenetic errors occurred before banding was available. Where sibs of those who were investigated in the pre-banding era request information as to their own risks of having an affected child, it is essential to reinvestigate the family using the best current techniques.

The family were originally under the care of Dr Leslie Scott, whom we thank for his help in the preparation of this report.

References

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Correspondence and requests for reprints to Dr G T McCarthy, Mac Keith Children's Centre, Royal Alexandra Hospital for Sick Children, Dyke Road, Brighton BN1 3JN.