Wellington Hospital, for assistance with the illustrations.

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References

- Howard-Peebles PN, Stoddard GR. Pericentric inversions of chromosome number 9: benign or harmful. *Hum Hered* 1979;**29**:111-7.
- Leonard C, Hazael-Massieux P, Bocquet L, Larget-Piet L, Boué J. Inversion pericentrique inv(2)(p11q13) dans les familles non apparentées. *Hum Genet* 1975;**48**:121-8.
- van der Linden AGJM, Pearson PL, van de Kamp JJP. Cytological assessment of meiotic exchange in a human male with a pericentric inversion of chromosome No 4. *Cytogenet Cell Genet* 1975;**14**:126-39.
- Teyssier JR, Bajolle F. Duplication-deficiency of chromosome 18, resulting from recombination of a paternal pericentric inversion, with a note for genetic counselling. *Hum Genet* 1980;**53**:195-200.
- Crawford Md'A, Mason MK. A pericentric inversion in three generations of a family. *Bull Eur Soc Hum Genet* 1973:46-8.
- Baccichetti C, Lenzini E, Peserico A, Tenconi R. Study on segregation and risk for abnormal offspring in carriers of pericentric inversion of the (p11q13) segment of chromosome 2. *Clin Genet* 1980;**18**:402-7.

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Partial deletion of the long arm of chromosome 4: a clinical syndrome

SUMMARY Partial deletion of the long arm of chromosome 4 at q31 results in a clinical syndrome of mental retardation, characteristic ears, facial bone hypoplasia, cleft palate very prone to scarring on repair, and specific hand abnormalities. A female, aged 9 years, is described and compared with six other reported cases.

Case report

The proband is the 9-year-old daughter of a 30-year-old father and 25-year-old mother. The parents are

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Argentinian emigrants of Spanish descent. There are two female sibs, aged 7 and 4, who are normal. There is no history of maternal or paternal radiation or miscarriage in the mother. The proband was delivered normally at term and had a birthweight of 3.1 kg. A cleft palate was noted at birth. She was described as a quiet baby, who needed to be woken for feeds. There was hard evidence of developmental retardation by the age of 1 year. Three attempts were made to repair the palate in Argentina at the age of 3, 5, and 6 years, all resulting in palatal fistula and scarring. Her speech is particularly retarded and indistinct, seemingly out of proportion to her general developmental level. There were only half a dozen distinguishable words and the child was frustrated by her inability to be understood.

CLINICAL EXAMINATION

Her height was 122 cm (10th centile), and her head circumference was 48 cm (less than the 3rd centile). She had prominent and small ears coming to a rounded point superiorly (fig 1). She had mid-facial and mandibular hypoplasia and a broad nasal bridge (fig 2), small 5th metacarpal bones and small distal phalanges of the 5th digits (fig 3). In addition there was some irregularity of the outline of the distal

FIG 1 Ear abnormality, pointed and prominent.

FIG 2 The proband. Note small jaw, facial bone hypoplasia, and prominent ears.

phalanx of the 4th digit on x-ray. She had a scarred palate with palatal fistula (fig 4). The palate moved in response to the gag reflex. There was no clinical abnormality in the cardiovascular system. Her teeth were within the normal limits.

IQ testing (WISC) was 57 though it was felt by the testing psychologist that this was not an accurate level because of her disproportionate problems with speech and lack of formal education. She particularly impressed as being more advanced in socialisation and comprehension than speech.

CYTOGENETIC INVESTIGATIONS

Initial analysis showed a partial deletion of the long arms of a B group chromosome. G banding by a modification of the method of Seabright¹ with subsequent staining with Leishman revealed a

FIG 3 Finger and hand abnormalities, small 5th metacarpal and distal phalanges.

FIG 4 Scarred palate repair with palatal fistula.

FIG 5 Trypsin-Leishman banding showing deletion of chromosome 4 at q31.

karyotype of 46,XX,del(4)(pter→q31) making the child monosomic for q31→qter (fig 5). Karyotypes of both parents were normal.

Discussion

Five previously reported cases and the present case are compared in the table. Like Townes *et al*,² we feel that, despite the paucity of reported cases, a definite syndrome appears to be emerging consisting of mental retardation, typical small prominent 'satyr' ears, cleft palate, very prone to scarring on repair, facial bone hypoplasia, oropharyngeal hypotonia resulting in airway and speech problems, and the peculiar abnormality of the 5th digit consisting of a hypoplastic distal phalanx. The clinical details of these cases are summarised in the table. Two of the reported cases died within hours of birth. One case is older, being 14 at the last report in 1972. Like most other deletion syndromes there is no parental age effect (with the possible exception of monosomy of the short arm of chromosome 18). There has been one case report resulting from a paternal translocation,² so cytogenetic studies are important for both prognosis and genetic counselling. Although Golbus *et al*³ and Ferrier and Freund⁴ G banded the

TABLE 4q— syndrome

	Present case	Townes <i>et al</i> ²	Golbus <i>et al</i> ³	Ferrier and Freund ⁴	Van Kempen ⁵	Frias <i>et al</i> ⁶
Chromosomes	del(4)(q31)	del(4)(q31)	4q—	4—	del(4)(q31)	del(4)(q31)
Sex	F	M	F	F	M	M
Age at report	9 years	Died 1 hr	6 months	4 months	14 years	3 months
Mental retardation	+	?	±	±	+	+
Satyr ears	+	+	+	—	+	+
Cleft palate	+	+	+	+	+	+
Facial bone hypoplasia	+	+	+	+	+	+
Oropharyngeal hypotonia	+	+	+	+	±	+
Cardiac defects	—	—	+	+	+	—
5th digit abnormality	+	+	—	+	+	+

chromosomes and identified the deleted chromosome as a number 4, they did not indicate the exact breakpoint. However, from the published karyotypes it appears to be at q31 as in the four other cases. Thus a definite chromosome abnormality, monosomy of 4q31→qter appears to be responsible for a clinical syndrome.

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References

- ¹ Seabright M. A rapid banding technique for human chromosomes. *Lancet* 1971;ii:971-2.

- ² Townes PL, White M, Marzo GV. 4q—syndrome. *Am J Dis Child* 1979;133:383-5.
- ³ Golbus MS, Conte FA, Daentl DL. Deletion from the long arm of chromosome 4 associated with congenital anomalies. *J Med Genet* 1973;10:83-5.
- ⁴ Ferrier S, Freund M. A propos d'un cas de deletion du bras long du chromosome B4 (B4q-). *Arch Genet* 1974;47:16-26.
- ⁵ Van Kempen C. A patient with congenital anomalies and a deletion of the long arm of chromosome 4 (46, XY,del(4)(q31). *J Med Genet* 1975;12:204-6.
- ⁶ Frias JL, Nelson RM, Ray SL. Deletion of the long arm of chromosome 4: a clinically identifiable syndrome? *Birth Defects* 1978;14:355-8.

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