Case reports

Wellington Hospital, for assistance with the illustrations.

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References


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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 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 Seabright with subsequent staining with Leishman revealed a 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, 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, opharyngeal hypoplasia 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, so cytogenetic studies are important for both prognosis and genetic counselling. Although Golbus et al and Ferrer and Freund G banded the

**TABLE 4q—syndrome**

<table>
<thead>
<tr>
<th>Chromosomes</th>
<th>Present case</th>
<th>Townes et al</th>
<th>Golbus et al</th>
<th>Ferrier and Freund</th>
<th>Van Kempen</th>
<th>Frias et al</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td>F</td>
<td>M</td>
<td>F</td>
<td>F</td>
<td>M</td>
<td>M</td>
</tr>
<tr>
<td>Age at report</td>
<td>9 years</td>
<td>Died 1 hr</td>
<td>6 months</td>
<td>4 months</td>
<td>14 years</td>
<td>3 months</td>
</tr>
<tr>
<td>Mental retardation</td>
<td>+</td>
<td>?</td>
<td>±</td>
<td>±</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Satyr ears</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Facial bone hypoplasia</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Oropharyngeal hypoplasia</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Cardiac defects</td>
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<td>+</td>
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<td>—</td>
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<tr>
<td>5th digit abnormality</td>
<td>+</td>
<td>+</td>
<td>—</td>
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</tbody>
</table>
chromosomes and identified the deleted chromosome as a number 4, they did not indicate the exact break-point. 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

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