The variability of the h regions can not be determined precisely when G bands are present. Consequently, this defeats the object of the exercise.

In our experience, the Q-C sequence is the most satisfactory and highly reliable (fig 2).

Familial X-linked mental retardation with an X chromosome abnormality and macro-orchidism

Sir,

Two forms of X-linked mental retardation have been described, one associated with an X chromosome which has a fragile site at Xq27 or 28, and the other associated with macro-orchidism. As a result of measuring the external genitals and calculating testicular volumes of retarded males with the fragile site at Xq27 or 28, Sutherland and Ashforth have suggested that these two forms of mental retardation are the same entity.

We have recently measured the genitals and calculated testicular volumes of some of the retarded males with fragile sites at Xq27 or 28 who were originally described by Harvey et al. Of the seven males examined (figure, table), six had testicular volumes greater than the 90th centile of Prader. Turner et al. have independently re-examined the chromosomes, under conditions appropriate for demonstration of fragile sites, of the males originally described with mental retardation and macro-orchidism. They found that these males do have the fragile site at Xq27 or 28. That finding, in conjunction with the report of Sutherland and Ashforth and the data presented here, confirm that the two forms of X-linked mental retardation recorded in published

References


FIG 2  Sequential Q- and C-banded metaphase showing enlarged secondary constriction (h) region of one chromosome 16 and complete inversion of one chromosome 9.
TABLE  Genital measurements of males with mental retardation and fra (X) (q27 or 28).

<table>
<thead>
<tr>
<th>Subject</th>
<th>Age</th>
<th>Penis Length</th>
<th>Circumference</th>
<th>Testicular volume (ml)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Right</td>
<td>Left</td>
<td></td>
</tr>
<tr>
<td>Family A</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>II.4</td>
<td>54</td>
<td>11.0</td>
<td>8.0</td>
<td>42</td>
</tr>
<tr>
<td>II.10</td>
<td>46</td>
<td>9.0</td>
<td>8.0</td>
<td>24</td>
</tr>
<tr>
<td>II.11</td>
<td>44</td>
<td>13.0</td>
<td>7.5</td>
<td>18</td>
</tr>
<tr>
<td>III.9</td>
<td>17</td>
<td>10.0</td>
<td>8.5</td>
<td>38</td>
</tr>
<tr>
<td>Family B</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>III.1</td>
<td>23</td>
<td>10.0</td>
<td>9.0</td>
<td>39</td>
</tr>
<tr>
<td>Subject TK</td>
<td>15</td>
<td>11.0</td>
<td>8.5</td>
<td>37</td>
</tr>
<tr>
<td>Subject NK</td>
<td>18</td>
<td>12.0</td>
<td>10.0</td>
<td>30</td>
</tr>
</tbody>
</table>

Family identification as in Harvey et al. Subjects TK and NK are brothers from another family mentioned in their addendum.

reports are the same. Hence, it is now possible to add large testes, as well as small ones, to the list of indications for chromosome studies.

Grant R Sutherland*, C G Judge†, and S Wiener‡

* Cytogenetics Unit, Adelaide Children's Hospital, North Adelaide, South Australia 5006; † Children's Cottages, Kew, Victoria 3101; and ‡ Chromosome Laboratory, St Nicholas Hospital, Carlton South, Victoria 3053, Australia

Correspondence

References


Covesdem syndrome

Sir,

I am writing to point out the marked similarity between the patients reported by Wadia et al.2 and those with the Robinow 'fetal face' syndrome.2

The features described in the patients reported by Wadia et al.1 namely hemivertebrae, rib defects, mesomelia, short stubby fingers, hypertelorism, depressed nasal bridge, and teeth anomalies are all features of the Robinow syndrome, as is the small penis reported in case 1. There also appears to be a marked overall facial similarity to the Robinow syndrome, although the article does not contain good close-up photographs.

The x-ray changes in the spine, ribs, and forearms are all entirely consistent with the Robinow syndrome; in particular, the changes at the elbow in fig 6 are almost identical to the changes shown in fig 4 in an article by Wadlington et al.4 which describes four cases of the Robinow syndrome. Although an autosomal dominant inheritance was suggested in the original description of the Robinow syndrome4 on the basis of two affected generations, Wadlington et al.5 described a sibship containing two affected subjects with normal parents and postulated autosomal recessive inheritance with occasional manifestation in the heterozygote. The mode of inheritance in the family reported by Wadia et al.1 would be consistent with this theory.

Correspondence

References