Adjacent 2 translocation involving 13q and 21q

SIR,

The article in Journal of Medical Genetics entitled ‘Adjacent 2 translocation involving 13q and 21q’ (1982;19:314–5) states that this case is the first involving chromosomes 13 and 21 with an adjacent 2 disjunction in the infant and a balanced reciprocal translocation involving the long arms of chromosomes 13 and 21 in the mother.

We have studied a female carrier of a translocation in which the long arms of chromosomes 13 and 21 were involved. Identification with G banding (GTG) was not conclusive enough to enable us to establish definite breakpoints but, together with R banding (RBA), would suggest the following karyotype: 46,XX,t(13;21)(q21;q21).

The offspring of this woman suggest that this translocation carries a high risk. The first child died just after delivery in another hospital without cytogenetic study. The second child had dysmorphic features with partial trisomy 13 and partial monosomy 21 owing to an adjacent 2 meiotic disjunction. His karyotype was 46,XY,−21,+der(13),t(13;21) (q21;q21). The third child had the phenotype of Down’s syndrome because of a 3:1 segregation and his karyotype was 47,XY,+21,t(13;21)(q21;q21).

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Reference

1 Prieto F, Badia L, Asensi F, Roques V. Two reciprocal translocations t(9p + ;13q−) and t(13q−;21q+). A study of the families. Hum Genet 1980;54:7–11.

Pyloric stenosis: children vs sibs

SIR,

We have reported1 findings in the relatives of patients with pyloric stenosis which showed, for female patients, more children affected than sibs. This is unexpected on a simple multifactorial threshold model and has led us and others to speculate whether there may be some direct maternal effect, though there is no indication, on the small series available, that maternal half-sibs are more often affected than paternal half-sibs.2 We have continued to follow the children of the female patients born between 1933 and 1949 (but not those born between 1921 and 1932, who are not likely to have further children) and the relatively high risk to children has now disappeared. The data on

Reference

the children at the time of the 1967 follow-up are summarised in table 1.

In the 1980 follow-up of the 179 women born in 1933 to 1949, 27 were not traced (25 were not traced through the National Health Service and two had emigrated). The information on the further children of the 152 who were traced is summarised in table 2.

Adding these new data, the totals for children are 21 sons affected in 173 (12·1%) and nine daughters affected in 182 (4·9%). These are now close to the figures for sibs for the same series of female patients, 10·0% and 6·3% for brothers and sisters respectively.

However, it will be seen from tables 1 and 2 that there is an indication of a secular change with a falling proportion of children affected among the more recently born, and there was also some suggestion of this in the figures for sibs. There are also indications that the incidence of pyloric stenosis in the general population may be falling. There is then a case for comparing contemporary births of sibs and children, rather than sibs and children of the same patients.

If we compare the children of patients born in 1933 to 1942 (mostly born between 1945 and 1975) and the sibs of patients born in 1950 to 1965 (who were also mostly born between 1945 and 1975) the figures are 7/51 for sons and 2/61 for daughters, and 8/103 for brothers and 0/105 for sisters. This suggests a higher risk for children. However, the numbers are small and a somewhat larger series from Belfast for patients born in 1957 to 1969 gave 20/160 brothers and 6/159 sisters, close to the figures for the children in the London series.

It is therefore probable that the proportion of sibs and children affected is similar and there is no need to postulate a direct maternal effect.

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References


Delineation of trisomy 9 syndrome

Sir,

In response to Frolich in the August 1982 issue of Journal of Medical Genetics (19:316–7) concerning delineation of trisomy 9 syndrome, we describe an infant who lived for 5½ days. Skin fibroblast and blood lymphocytes showed trisomy 9, with one of the 9 homologues exhibiting an extra band in the 9qh+ region. (The 9qh+ variant was not in duplicate.) Variants in association with trisomy 9 have been previously observed. In the report in the October 1981 issue of Journal of Medical Genetics (18:390–2) by Frydman et al, the partial inversion of 9 described might well be reassessed in the light of recent studies, although such variants remain excellent markers, as was shown in the aforementioned report.

This baby was born to a mother who already had one normal child. The pregnancy had been unremarkable and she had only taken Amoxil and Debendox. At delivery there was a suggestion of polyhydramnios which had not been appreciated before. Birth weight was 2170 g. At birth the baby was mildly depressed, with Apgars of 4 at 1 minute, 6 at 5 minutes, and 9 at 10 minutes. Because of orofacial abnormalities the baby was unable to be intubated; however, he was easily resuscitated with bag and mask. There were a number of physical abnormalities. The head was very small with a circumference of 20 cm, well below the 10th centile for 38 weeks. There was a bilateral cleft lip and palate and the philtrum was on the tip of the nose, which was broad. The centre of the forehead protruded. Hypertelorism was present and palpebral fissures were narrow. The anterior and posterior fontanelles were both very large, but tension

TABLE 1 1967 follow-up.

<table>
<thead>
<tr>
<th>Year of birth</th>
<th>Number</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td>1920–1932</td>
<td>46</td>
<td>40 (9R)</td>
<td>36 (5R)</td>
</tr>
<tr>
<td>1933–1942</td>
<td>64</td>
<td>42 (7R)</td>
<td>50 (2R)</td>
</tr>
<tr>
<td>1943–1949</td>
<td>115</td>
<td>106 (20A)</td>
<td>100 (7A)</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>106 (20A)</td>
<td>100 (7A)</td>
</tr>
</tbody>
</table>

R = Rammstedt, T = tumour felt and medical treatment, A = affected

TABLE 2 1980 follow-up of further births.

<table>
<thead>
<tr>
<th>Year of birth</th>
<th>Number</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td>1933–1942</td>
<td>55</td>
<td>9</td>
<td>11</td>
</tr>
<tr>
<td>1943–1949</td>
<td>97</td>
<td>58 (1R)</td>
<td>71 (2R)</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>67 (1R)</td>
<td>82 (2R)</td>
</tr>
</tbody>
</table>
Pyloric stenosis: children vs sibs.

C O Carter, V Hickman and K Evans

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