Correspondence

Higher risk to D;G translocation carriers of tdic(13;21) as compared to tdic(14;21)

Sir,

The generally quoted risk for maternal carriers of t(Dq21q) to produce trisomy 21 offspring has been approximately 10%.1,2 This figure has been determined largely from data of families carrying t(14q21q). There are only a few reports3-5 of t(13q21q) families, and in one sample of 33 families in which a 10% maternal recurrence risk was determined,3 there were no t(13q21q) detected. A similar paucity of t(13q21q) has been reported in the consecutive newborn data of 59,452 infants summarised by Jacobs.6 In that data six (14q21q), one each of t(15q21q), t(13q22q), t(14q22q), and t(15q22q), but no t(13q21q) were found. In this laboratory six familial t(14q21q) and four familial t(13q21q) have been ascertained through translocation trisomy 21 offspring. Whereas the t(13q21q) may be over-represented in this sample, this has permitted a preliminary comparison of segregation patterns (table) in the two translocation types. The data have been compiled by the removal of probands, parental carriers of probands, and carriers linking generations as traced back from the proband, after the method of Stene.1

In the present data the risk for maternal carriers of t(14q21q) is as reported previously.1,2 However, the maternal risk for t(13q21q) is significantly higher ($\chi^2 = 5.4; p = 0.05$) at approximately 50%. Also the spontaneous abortion rate in maternal carriers of either translocation is increased at 30% of recognised conceptuses, as compared with 5-3% for pooled male carriers. This difference is also significant ($\chi^2 = 11.9; p = 0.001$). Such findings have not been reported previously and it would be interesting to examine the pooled data of several laboratories with single or small numbers of families. In this connection we would be interested in corresponding with other laboratories.

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References


Cause of neural tube defects

Sir,

James1 suggested that there are two sorts of causes of anencephaly. One is definitely environmental, affecting predominantly female embryos, the other may be either environmental or genetic, but it seems to affect the sexes equally. Whatever causes anencephaly probably can be expected to cause spina bifida too. James went on to suggest that his hypothesis carried implications for any clinical trials aimed at identifying the environmental agent. He recommended that in any study aimed at

<table>
<thead>
<tr>
<th>Translocation</th>
<th>Sex of carrier</th>
<th>No of families</th>
<th>No of sibships</th>
<th>Carriers</th>
<th>Normal</th>
<th>Trisomy*</th>
<th>Spontaneous abortions</th>
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</thead>
<tbody>
<tr>
<td>tdic(14;21)</td>
<td>F</td>
<td>6</td>
<td>9</td>
<td>13</td>
<td>5</td>
<td>2 (10.0%)</td>
<td>10</td>
</tr>
<tr>
<td></td>
<td>M</td>
<td>7</td>
<td>9-5</td>
<td>6-5</td>
<td>1 (0.8%)</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>tdic(13;21)</td>
<td>F</td>
<td>6</td>
<td>0</td>
<td>5-5</td>
<td>5 (50%)</td>
<td>5</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>M</td>
<td>6</td>
<td>8-5</td>
<td>8-5</td>
<td>2 (10.5%)</td>
<td>2</td>
<td></td>
</tr>
</tbody>
</table>

*Not including spontaneous abortions in calculation.
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