Meiotic behaviour of two human reciprocal translocations

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SUMMARY The meiotic behaviour of two male human reciprocal translocations is described. One patient had an unbalanced son and a chain configuration. The second had a stillborn child and a ring corresponding to an adjacent I segregation. The meiotic behaviour of chromosomal rearrangements must be investigated for proper genetic counselling.

According to the data of Jacobs et al the incidence of balanced chromosomal rearrangements in the adult population is 3.32 per 1000. The most common type is the translocation. In newborns the incidence of balanced reciprocal translocations is less than 1 in 1000, but in subfertility clinics the incidence is much higher. In our series of 1015 subfertile males (up to 31 May 1980) in whom mitotic or meiotic studies or both have been carried out, we have found five reciprocal translocations. In spite of the prognostic importance of the meiotic behaviour of the translocation products, to our knowledge only ten cases have been published. In this paper we describe the meiotic findings in two subfertile males, heterozygous for reciprocal translocations.

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Material and methods

The two cases were referred for chromosome studies by the Department of Andrology, Fundación Puigvert. Peripheral blood cultures were carried out using commercial kits and analysed by G banding and Q banding. In case 1 meiosis was studied in semen using the technique of Sperling and Kaden as modified by us. In case 2 a testicular biopsy was obtained under local anaesthesia and meiotic preparations were made using the technique of Evans et al.

Results

CASE 1. 46,XY,t(10;13)(q25;p12)
This patient, aged 34, had a son with mental

FIG 1 Diakinesis and chain configurations in the carrier of a (10;13) translocation.
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<table>
<thead>
<tr>
<th>Reference</th>
<th>Translocation</th>
<th>Meiotic configuration</th>
<th>Abnormal sperms (million/ml)</th>
<th>IH + II (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>46,XY,t(5;7)(q13;q11)</td>
<td>Normal son and daughter with malformations</td>
<td>0.8</td>
<td>38</td>
</tr>
<tr>
<td>2</td>
<td>46,XY,t(5;7)(q13;q11)</td>
<td>Normal son and daughter with malformations</td>
<td>7.5</td>
<td>23</td>
</tr>
<tr>
<td>3</td>
<td>46,XY,t(5;7)(q13;q11)</td>
<td>Aneuploid spermata</td>
<td>39</td>
<td>5</td>
</tr>
<tr>
<td>4</td>
<td>46,XY,t(5;7)(q13;q11)</td>
<td>Aneuploid spermata</td>
<td>4.5</td>
<td>31</td>
</tr>
</tbody>
</table>

CASE 2. 46,XY,t(5;7)(q13;q11)

This patient, aged 25, requested counselling because his wife had given birth to a malformed fetus at 8 months' gestation. Cytogenetic studies showed a balanced (5;7) reciprocal translocation.

The spermiogram showed oligozoospermia. At meiosis, the number of divisions was low, but all 19 diakineses studied had a tetravalent in the form of an open chain resulting from failure of chiasma formation in one of the pairing regions of the translocation cross (fig 1).

Discussion

The number of meiotic studies in reciprocal translocation carriers is still low (table) and none of the published articles deals in depth with the genetic counselling implications of the meiotic figures observed, although Chandley et al have repeatedly indicated that chain configurations carry a worse prognosis than rings, because of the fact that in such cases meiotic breakdown is more frequent.

Case 1 had a translocation involving band 10q25, which has recently been shown to be a fragile site, although it is only expressed by BrdU incorporation. In Robertsonian fusions, counsellors may give a 33% theoretical risk or a 2 to 10% empirical risk depending on whether the carrier is the male or the female. However, as Moses et al have shown in Lemur hybrids, this risk may be as low as 0% if pairing of the trivalent always takes place in the cis configuration, and higher than the theoretical 33% if pairing is in trans.

In reciprocal translocations, the theoretical risk is at least 50%, because adjacent segregation is probably as frequent as alternate segregation. However, in most published cases adjacent segregation has been the rule.

Finally, it must be taken into account that some types of translocation predispose to 3:1 disjunction. These are the translocations involving an acrocentric, particularly a short one, and those with extreme disparity in total chromosome length. The behaviour of human reciprocal translocations has recently been reviewed by Jalbert et al.
Thus, the study of the meiotic behaviour of translocated chromosomes and of their pairing characteristics should be considered essential for counselling translocation carriers. Although it is not always easy to obtain gonadal material, the possibility of carrying out meiotic and synaptoneomal complex studies in the ejaculate enables investigations in all male carriers to be carried out.

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References


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