Incidence of Chromosomal Rearrangements in Couples with Reproductive Loss

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Summary. We report on 50 couples with reproductive loss who did not have any detectable chromosome abnormality. A history of a previous child with multiple congenital abnormalities may be significant in identifying couples with a structural rearrangement. Only by studying more families can this hypothesis be tested. Studies of abortus tissue reveal a high percentage of chromosome abnormalities but a very low incidence of unbalanced translocations. Cytogenetic studies are indicated in a couple which has a past history of spontaneous abortions and a previous child with multiple congenital anomalies.

Introduction

Chromosome analysis is frequently recommended for couples which have had two or more episodes of reproductive loss. Reproductive loss includes first trimester spontaneous abortions and stillborn or liveborn infants with multiple congenital anomalies. The incidence of chromosome abnormalities in this group of patients ranges from 1/10 (Kim et al., 1975) to 1/30 (Tsenghi et al., 1976). The higher incidence figures are attributed to the more exact identification of balanced chromosome rearrangements using the newer banding techniques (Kim et al., 1975).

The frequency of balanced chromosome translocations in the normal population is believed to be about 1/500. Jacobs (1977) reports 104 reciprocal translocations in a combined series of 54,749 newborn infants. The results of cytogenetic studies in couples with reproductive loss are significant compared with those in the general population. However, the variability in these studies poses several questions.

Because of the different incidence figures of chromosome abnormalities, we decided to study a group of 50 couples with a history of reproductive loss. We compare our findings with the relative incidence of chromosomal rearrangements detected in a group of patients who were referred for amniocentesis. Banding techniques are routinely utilized in our laboratory.

Materials and Methods

From September 1975 to December 1978, 50 couples with the problem of reproductive loss were referred for cytogenetic studies. Each member received complete endocrinologic and gynecologic evaluations and no abnormalities were found.

Cultures of peripheral blood were harvested after 72 h and all slides were stained using the G-banding (GTG) technique (Klinger, 1972). Twenty metaphases were analyzed and four karyotypes were prepared from each culture.

Results

Analysis of banded karyotypes from the 50 couples studied revealed no chromosome abnormalities. The majority of couples (40/50) were referred because of two or more consecutive first trimester abortions. Ten couples had a stillborn delivery and of this group only three of the stillborn infants had multiple congenital anomalies (Table 1).

While this study was in process, three balanced translocations were discovered in patients who had amniocentesis (Figs. 1–3). A total of 788 amniotic fluid specimens were analyzed.
Table 1. Reason for referral

<table>
<thead>
<tr>
<th>Reason for referral</th>
<th>Count</th>
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<tbody>
<tr>
<td>Two first trimester spontaneous abortions</td>
<td>13</td>
</tr>
<tr>
<td>More than two first trimester spontaneous abortions</td>
<td>27</td>
</tr>
<tr>
<td>Spontaneous abortion + stillborn (normal phenotype)</td>
<td>7</td>
</tr>
<tr>
<td>Spontaneous abortion + stillborn (multiple congenital abnormalities)</td>
<td>3</td>
</tr>
<tr>
<td></td>
<td>50</td>
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Discussion

Cytogenetic studies are routinely included in the evaluation of couples who have had two or more episodes of reproductive loss. In our 50 couples with similar reproductive histories, no chromosomal rearrangements were found. These results lead us to question the incidence of reciprocal translocations in this group of patients and to speculate as to what historical findings...