The paper aims to identify the type and frequency of chromosomal abnormalities in couples with malformed stillbirths or infants deceased, as well as to identify the particularities in such couples. The study was accomplished on 32 couples who had malformed stillbirths or infants deceased. Both members of the couples were investigated through medical and familial anamnesis and conventional cytogenetic analyses. For the identification of couple’s characteristics with chromosomal abnormalities, bivariate analysis was realized. In this group of patients, the overall frequency of chromosomal abnormalities was of 4.68%. The balanced chromosomal abnormalities identified were: reciprocal (n=2) and Robertsonian translocations (n=1). Bivariate analyses revealed the statistical significant differences in male and female age, presence of spontaneous abortion and familial anamnesis positive for reproduction failure in the couples with chromosomal abnormalities.

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In order to establish the significance between the variables that characterized the couples with chromosomal abnormalities and the couples with normal karyotype, we realized bivariate analyses. The t-test and χ²-test were used for statistical evaluation. The level of significance was p<0.05.

RESULTS

From the 32 couples, 15 present in their reproductive antecedents, stillbirth malformed child and 17 couples present malformed infants deceased.

- The characteristics of couples included in our study were:
  - Maternal ages vary between 20 and 38 years old, with a mean age of 28.88 years.
  - Paternal ages vary between 23 and 38 years old, with a mean age of 30.38 years.
  - Reproductive history revealed the presence of spontaneous abortions in 47% of couples, in which the number varies between 1-5 abortions. Only 20% of couples have spontaneous abortion occurred in the first trimester of pregnancy.
  - Family inquires revealed the existence of other cases of reproductive failure in the family for 31% of couples (spontaneous abortion 19%, children with multiple congenital abnormalities syndromes 9%, primary sterility 3%).
  - The malformed stillbirths or infants were diagnosed with Down syndrome (25%), Patau syndrome (19%), Edwards syndrome (6%), VACTER malformative association (9%) and other plurimalformative syndromes (41%). Cytogenetic confirmation of syndrome was accomplished in 56% of cases.
  - Physical examination of both partners of the couples revealed no particularities. Body mass index (BMI) for the male partner of a couple who presented two recurrent abortions in 1st trimester of pregnancy.

- The translocation t(11;18)(p15;p11) was identified in the female partner of a couple who presented two spontaneous abortions and a female infant with Down syndrome.

- The translocation t(4;8)(p16;p23) was identified in the male partner of a couple who presented two recurrent abortions in first trimester of pregnancy and a female stillbirth with phenotype suggestive for trisomy 18, due to possible translocation segregation of 3:1.

- The translocation t(13q;21q) was identified in the female partner of a couple who presented two spontaneous abortions and a female infant with Down syndrome.

Bivariate analyses made on the couples with chromosomal abnormalities and couples with normal karyotype (table no. 2) revealed the presence of statistical significant differences regarding the following variables:

- female’s and male partner’s ages were younger in the couples with chromosomal abnormalities
- the average number of spontaneous abortions was greater in couples with translocations as against in couples with normal karyotype
- family inquiry revealed the presence of other cases of reproduction failure in family for 100% of couples with translocations and for 24% of couples with normal karyotype.

Table no. 2. Bivariate analyses in couples with and without chromosomal abnormalities

<table>
<thead>
<tr>
<th>Chromosomal abnormalities in couples with malformed stillbirth or infants</th>
<th>No</th>
<th>Yes</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Maternal age (years)</td>
<td>29±3,86</td>
<td>24,67±0,58</td>
<td>0,049</td>
</tr>
<tr>
<td>Paternal age (years)</td>
<td>30,79±3,65</td>
<td>26,33±1,53</td>
<td>0,047</td>
</tr>
<tr>
<td>No of spontaneous abortions</td>
<td>0,79±1,29</td>
<td>2,67±1,25</td>
<td>0,022</td>
</tr>
<tr>
<td>Maternal weight (kg)</td>
<td>59,17±4,92</td>
<td>55,33±5,83</td>
<td>0,214</td>
</tr>
<tr>
<td>Maternal height (m)</td>
<td>1,64±0,05</td>
<td>1,68±0,04</td>
<td>0,119</td>
</tr>
<tr>
<td>Maternal BMI (kg/m²)</td>
<td>22,18±2,26</td>
<td>19,56±1,21</td>
<td>0,06</td>
</tr>
<tr>
<td>Paternal height (m)</td>
<td>1,79±0,05</td>
<td>1,81±0,02</td>
<td>0,601</td>
</tr>
<tr>
<td>Paternal weight (kg)</td>
<td>86,90±18,6</td>
<td>82±5,29</td>
<td>0,657</td>
</tr>
<tr>
<td>Paternal BMI (kg/m²)</td>
<td>18,49±1,84</td>
<td>16,98±2,13</td>
<td>0,190</td>
</tr>
<tr>
<td>Positive family inquiry</td>
<td>24% (29)</td>
<td>100% (3)</td>
<td>0,040</td>
</tr>
<tr>
<td>Oral contraceptive</td>
<td>31% (9)</td>
<td>33% (1)</td>
<td>0,560</td>
</tr>
<tr>
<td>Existence of healthy child</td>
<td>38% (11)</td>
<td>0</td>
<td>0,496</td>
</tr>
<tr>
<td>Pregnancy age (weeks)</td>
<td>35,52±3,43</td>
<td>38±2</td>
<td>0,232</td>
</tr>
</tbody>
</table>

DISCUSSION

The literature data notify that maternal fatness in pregnancy is one of the frequent factors involved in the etiology of stillbirth. In our group of patients, we excluded this factor by BMI determination which revealed that majority of our female patients (91%) had a normal weight, 6% were overweight and 3% were underweight.

Cytogenetic analyses revealed the presence of balanced chromosomal abnormalities in one member of a couple with an overall frequency of 4.68%, frequency similar to the ones depicted in other studies.(4,5)
The female/male ratio observed in our study is almost similar to the ratio reported in most of the reported studies. The predominance of chromosomal abnormalities in females appears to be due to the fact that chromosomal abnormalities that are compatible with fertility in females may be associated with sterility in males.

The type of chromosomal abnormalities detected in our study is similar to the type reported in other study. The statistical significant differences regarding the ages of couple partners with chromosomal abnormality emphasize the role of chromosomal abnormalities in the production of genetic abnormal gametes in relatively young ages, which give rise to foetus with chromosomal pluriformative syndromes. According to the segregation of chromosomal abnormalities in embryo, the anomaly is either compatible with survival and gives rise to a malformed child, or is incompatible with life or gives rise to spontaneous abortion.

Family inquiry positive for reproduction failure in the family of couples with chromosomal abnormalities could signify the transmission of derivative chromosome in balanced state, suggesting for the necessity of cytogenetic investigations of the family.

CONCLUSIONS

Parental balanced chromosomal rearrangements are important factors in the etiology of malformed stillbirth or infants.

In the couples with malformed stillbirth or infants, the probability that one member could be the carrier of balanced chromosomal abnormalities is increased by the presence of spontaneous abortions, age of couple’s members younger than 30 years old and family inquiry positive for reproductive failure.

REFERENCES