Abstract:

Introduction:

Definition of Recurrent pregnancy loss is having 3 or more miscarriages. After two or three miscarriages, a thorough physical examination and testing are recommended. In a small number of couples who have repeated miscarriages, one partner has chromosomal abnormalities. The purpose of this study is to determine the prevalence of these abnormalities in couples refer to our perinatal clinics at Imam Khomeini Hospital in Ahvaz.

Material and method:

This study was done on 320 couples who visited the perinatal clinic at Ahvaz's Imam Khomeini Hospital in a 2-year period, from 2021/03 to 2023/03, and had multiple abortions (more than three). The patients were initially evaluated, and a complete medical history was recorded. In order to assessment anatomical abnormalities vaginal sonography was used, along with antiphospholipid antibody syndrome testing and sperm analysis also was done. If the outcomes were not positive, in 142 cases both the husband and wife underwent karyotyping using the G-banding or CGH Array method, and the data were input into the software for Statistical analysis.

Result:

142 couples (284 people) were evaluated in this survey, 5 people with chromosomal abnormalities were detected (3.52% of couples) (1.76% people), 2.11% of females (3 females) in the ages ranged 18 to 45(mean 30.6), and 1.4% of men (2 men) in the range of 21 to 53 (mean:35.3).
Conclusion:

The prevalence of chromosomal abnormalities in our study was 3.52%, which of course is where we didn’t have abnormal initial findings. It seems performing chromosomal analysis in couples suffering repeated miscarriages should be in secondary priority, and first, the analysis in terms of anatomical and hormonal problems and immunological factors should be done.

Keywords:

recurrent abortion, chromosomal abnormalities, translocation.

Introduction:

Having a history of 3 or more abortions is called Recurrent pregnancy loss. Reasonable cause established in About 50% of couples with recurrent pregnancy loss, and There are disagreements about the causes of having PRL in some couples. uterine abnormalities, chromosomal abnormalities, autoimmune disorders, infectious diseases, coagulopathy disorders, endocrine disorders, and environmental factors are known as common causes of recurrent miscarriage. A complete clinical evaluation is necessary for couples which experiencing Recurrent pregnancy loss. The frequency and distribution of chromosomal abnormalities and the impact of parental chromosomal aberration on the pregnancy outcomes of couples with recurrent pregnancy loss remains controversial. According to Cytogenetic Studies of 608 couples with Recurrent Spontaneous Abortions in Northeastern Iran by Narjes Soltani et al.43 out of 1216 (620 couples) patients which had more than 2 abortions, had chromosome abnormalities in their karyotype. (1) Rola F. Turki, Mourad Assidi, et al. have cited in Associations of recurrent miscarriages with chromosomal abnormalities, thrombophilia allelic polymorphisms and/or consanguinity in Saudi Arabia A high rate of translocations (46 %) was associated to increased incidence of RPL. A significant correlation between consanguineous Recurrent pregnancy loss patients and chromosomal abnormalities (P < 0.05) was found(2). In Cytogenetic analysis in couples with recurrent pregnancy loss published by Serenat YALÇIN et al. Of 506 cases, chromosomal abnormalities were present in 15 (2.9%). Women were more frequently affected than men, with prevalence of 1.9% and 0.98%, respectively. Eight of the 15 cases (53.3%) showed structural deviations and 2 (13.3%) had numerical abnormalities. Additionally, 5 (33.3%) individuals were found to have chromosome variants. (3). The study
attempts to summarize the frequency of abnormal chromosomal karyotype couples, the
topography of abnormal types, and the frequency of the male and female carriers in recurrent
pregnancy loss.

method:
A study was done on 320 couples who visited the perinatal clinic at Ahvaz's Imam Khomeini
Hospital in 2 year period, from 2021/03 to 2023/03, and had multiple abortions (more than
three). The patients were initially evaluated, and a complete medical history was recorded. In
order to assessment anatomical abnormalities vaginal sonography was used, along with
antiphospholipid antibody syndrome testing and sperm analysis also was done. If the outcomes
were not positive, in 142 cases both the husband and wife underwent karyotyping using the G-
banding or CGH Array method, and the data were input into the software for Statistical analysis.

Results:
In this article, we study couples with a history of 3 or more than 3 abortions that visited the
Perinatal clinic at Ahvaz's Imam Khomeini Hospital in a 2-year period from 2021/03 to 2023/03.

142 couples (284 people) were evaluated in this survey, 5 people with chromosomal
abnormalities were detected (3.52% of couples) (1.76%people), 2.11% of females (3 females) in
the ages ranged 18 to 45(mean 30.6), and 1.4% of men (2 men) in the range of 21 to 53
(mean:35.3).

Different types of chromosomal abnormalities were discovered among the patients, the most
detected abnormalities were translocation, approximately 1.05% of 284 people have
translocation disorder, 2 female patients out of 142 female patients (1.40%) have different types
of translocation abnormalities (both balanced and Robertsonian translocation), one male
patient with translocation (balanced and nonreciprocal translocation) were detected (0.70%
males). Two inversions were found in abnormal karyotypes, one in female (0.70% females) and one in male patients (0.70% males).

Frequency Table

<p>| Karyotype F. |</p>
<table>
<thead>
<tr>
<th></th>
<th>Frequency</th>
<th>Percent</th>
<th>Valid Percent</th>
<th>Cumulative Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Valid</td>
<td>Normal</td>
<td>139</td>
<td>97.9</td>
<td>97.9</td>
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<tr>
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<td>Abnormal</td>
<td>3</td>
<td>2.1</td>
<td>100.0</td>
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<tr>
<td></td>
<td>Total</td>
<td>142</td>
<td>100.0</td>
<td>100.0</td>
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<table>
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<th>Frequency</th>
<th>Percent</th>
<th>Valid Percent</th>
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<tr>
<td>Valid</td>
<td>Normal</td>
<td>140</td>
<td>98.6</td>
<td>98.6</td>
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<tr>
<td></td>
<td>Abnormal</td>
<td>2</td>
<td>1.4</td>
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<td></td>
<td>Total</td>
<td>142</td>
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<table>
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<tr>
<th>Abortions</th>
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<th>Female</th>
<th>Total</th>
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</thead>
<tbody>
<tr>
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<td>2</td>
<td>3</td>
</tr>
<tr>
<td>Inversion</td>
<td>1</td>
<td>1</td>
<td>2</td>
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</tbody>
</table>
Discussion:

Chromosome abnormalities were found in 43 patients (3.54%), including 25 females and 18 males. Polymorphic variants were observed in 22 individuals. Structural chromosomal abnormality was detected in 40 cases, including balanced translocations (25 cases), Robertsonian translocations (4 cases), inversions (10 cases), and numerical chromosome aberrations (3 cases). (1) which was in the northeast of Iran and in corresponds with the 3.52% found in 284 patients in Ahwaz.

Data analysis showed that 7.6 % of patients had numerical or structural chromosomal abnormalities. A high rate of translocations (46 %) was associated with an increased incidence of RPL. A significant correlation between consanguineous RPL patients and chromosomal abnormalities (P < 0.05) was found. Both Factor V Leiden and Prothrombin A20210G allelic polymorphisms were significantly associated with a higher prevalence of RPL(2)

Of 506 cases that were assesse by YALÇIN S, ÖZTÜRK KH, ÖZBAŞ H, and Hilmi O , chromosomal abnormalities were present in 15 (2.9%). Women were more frequently affected than men, with prevalences of 1.9% and 0.98%, respectively. Eight of the 15 cases (53.3%) showed structural deviations and 2 (13.3%) had numerical abnormalities. Additionally, 5 (33.3%) individuals were found to have chromosome variants. (3)
In Asoke K. Pal's study, Out of 172 couples, 17 couples (9.88%) had different types of structural or numerical chromosomal abnormalities. Out of 17 couples, 8 (47.05%) had balanced translocations, 2 (11.76%) had the Robertsonian translocation, 5 (29.41%) had the pericentric inversion of chromosomes 8, 9, and Y, and only 2 (11.76%) women showed sex chromosome numerical aberrations. The percentages of chromosomal abnormalities in this study is about 2.8 times more than in our study in Ahvaz. In the study, we detected 40% of all abnormalities were balanced & Robertsonian translocation (1.4% of 142 couples). 20% were balanced & nonreciprocal translocation (0.70% couples), and 40% inversion (1.4% couples). (4)

In Le S and co-worker's study, Chromosome aberration was detected in 121 (3.74%) among 3235 RPL couples which included 75 female and 46 male cases at an individual level. 101 cases were structural aberrations including balanced translocations in 46 (38.0%) cases, Robertsonian translocations in 13 (10.7%) cases, inversions in 42 (34.7%) cases and 20 (16.5%) cases were numerical aberrations. 121 carriers and 428 non-carriers were followed up for two years, 55 carriers and 229 non-carriers were subsequent pregnant after diagnosis by natural conception or intrauterine insemination. (5)

In the study that published in 2018 in Ardabil, Iran 350 patients with at least two spontaneous abortions were analyzed. Parental chromosomal abnormalities were found in 18 cases (10.28% of couples). The percentage of parental chromosomal abnormalities in this study is three times higher than in our study in Ahvaz, Iran. (6)

In a Moroccan study published in 2018, that involved 238 couples with repeated abortions. the result was not significantly different from that reported worldwide, Chromosomal abnormalities were detected in 13 (6.1%) of 238 couples. Twelve of the chromosomal abnormalities were structural and one of them was numerical. In our study, chromosomal abnormalities were detected in 3.52% of couples, and all of the abnormalities were structural. (7)

In Serapinas. D et al study The chromosomal aberrations rate in Lithuania that may affect RM or the inability to conceive is 3%. The balanced translocations and mosaic aberrations were the most frequent. Chromosomal aberrations were more common among couples unable to conceive (p < 0.05) when compared to other groups of couples (with miscarriages). In total, 8.1% of the cases suffered from miscarriages in the family. There was no significant difference regarding whether more frequent miscarriages (p > 0.05) occurred on the male or female side. (8)

In the study that published in 2006 in Ahvaz by Malekasgar AM and his co-worker's Chromosomal analysis of these patients revealed three abnormal karyotypes (3.8%) in three women and two abnormal karyotypes in conceptions. Two of these couples had consanguineous marriage and the remaining women included one isochromosome for X [46, xX],
(xq)], two translocations [45, xx, t (15:21)] and [46, xx, t (7:14)], one trisomy ‘21’ (47, xx, +21), and a ring chromosome (46, xx, r(X). In addition, 27 conceptions had been reported for these five couples. These included 23 abortions with 18 of them within first trimester (78.26%) and four of them had abortions within second trimester (21.74%), one had a normal child, three had abnormal children, and one with stillbirth.(9)

In one retrospective study that was conducted at Balcalı Hospital in Adana region of Turkey, karyotype results of 1510 couples with a history of recurrent spontaneous abortion were evaluated. The chromosome aberrations were detected in 62 couples (4.1% of all couples). At an individual level, chromosome aberrations were found in a total of 65 cases (41 females and 24 male cases), with structural chromosomal aberrations in 58 cases including balanced translocations in 30 cases, Robertsonian translocations in 12 cases, deletions in seven cases, inversions in nine cases and numerical chromosome aberrations in seven cases. The results of the study indicated that structural aberrations, particularly translocations, were the most common type of aberration observed among couples who had experienced recurrent. (10)

In Franssen MT study that conducted in Netherlands Couples whose carrier status was ascertained after two or more miscarriages have a low risk of viable offspring with unbalanced chromosomal abnormalities. Their chances of having a healthy child are as high as non-carrier couples, despite a higher risk of miscarriage. (11)

In the article that published by Elghezal H in Tunisia Ninety-seven chromosome anomalies were detected among the 1,400 studied couples (6.93%). This prevalence appears to be in the range of previously reported studies in other populations but its two times more than our study in Ahvaz. (12)

Out of 795 couples, 28 (3.52%) were found to have a chromosomal abnormality (carrier group). Over half (65.5%) of the chromosomal abnormalities were balanced reciprocal translocations.(13)

In these studies, overall the probability of abnormal karyotype has been between 3to10 percent, which of course depends on the selection of the statistical population.

Conclusion:

The prevalence of chromosomal abnormalities in our study was 3,52%, which of course in where we didn’t have abnormal initial findings. It seems performing chromosomal analysis in couples suffering repeated miscarriages should be in secondary priority, and first, the analysis in terms of anatomical and hormonal problems and immunological factors should be done.
References:


9. Malekasgar AM, Motlagh ME, HASHEMI TM, Ghafari MA. Chromosomal Analysis Of Couples With Bad Obstetric Histoty. 2006


