



ISSN Print: 2664-8881
ISSN Online: 2664-889X
Impact Factor: RJIF 8
IJMS 2023; 5(2): 26-29
www.medicinejournal.in
Received: 18-06-2023
Accepted: 24-07-2023

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International Journal of **Medicine Sciences**

Analysis of chromosomal abnormalities pattern in couples with recurrent pregnancy loss in Khuzestan Province in 1400 and 1401(from 2021/3 to 2023/3)

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DOI: <https://doi.org/10.33545/26648881.2023.v5.i2a.41>

Abstract

Introduction: Recurrent pregnancy loss is characterized by 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. We assessed the prevalence of these abnormalities in couples referred to our perinatal clinics at Imam 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 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. A definite cause is established in no over 50% of couples, and many of the alleged causes of recurrent pregnancy loss are polemical. 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 with recurrent pregnancy loss. There is no consensus on the distribution and frequency of chromosomal abnormalities as well as the effect of parental chromosomal aberration on the pregnancy outcomes in couples who have recurrent pregnancy loss. Narjes Soltani *et al.* conducted Cytogenetic Studies on 608 couples who had Recurrent Spontaneous Abortions in Northeastern Iran and 43 out of 1216 (620 couples) patients who 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, consanguinity, and/or thrombophilia allelic polymorphisms in Saudi Arabia there was an association between increased rate of translocations (46%) and elevated RPL incidence. Chromosomal abnormalities and consanguineous Recurrent pregnancy loss were significantly correlated ($p < 0.05$) ^[2]. In a Cytogenetic study on couples who had recurrent pregnancy loss by Serenat YALÇIN *et al.* 15 (2.9%) out of 506 cases showed chromosomal abnormalities. Females were more affected compared to males (respectively 1.9% and 0.98%). Eight out of 15 cases (53.3%) reported structural deviations and two cases (13.3%) reported numerical abnormalities.

Five individuals (33.3%) had chromosome variants [3]. We summarized the topography of abnormal types, the rate of couples with abnormal chromosomal karyotype, and the frequency of the female and male 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 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 assess 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 studied couples who had 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% male patients). Two inversions were found in abnormal karyotypes, one in female (0.70% females) and one in male patients (0.70% males).

Frequency Table

Table 1: Result of female karyotype test

Karyotype F.					
	Frequency		Percent	Valid Percent	Cumulative Percent
Valid	Normal	139	97.9	97.9	97.9
	Abnormal	3	2.1	2.1	100.0
	Total	142	100.0	100.0	

Table 2: Result of male karyotype test

	Frequency	Percent	Valid Percent	Cumulative Percent
Valid	Normal	140	98.6	98.6
	Abnormal	2	1.4	100.0
	Total	142	100.0	

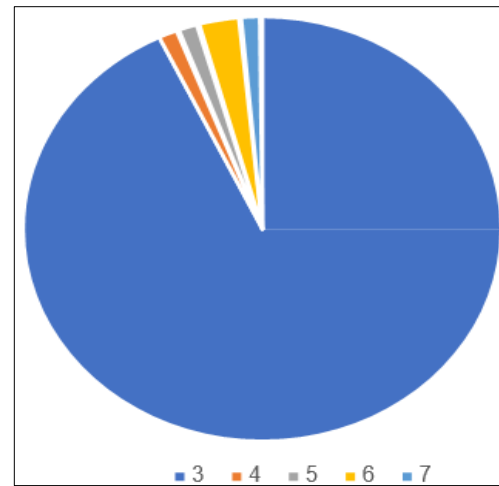


Fig 1: Number of previous abortions

Table 3: Percentages of each chromosomal abnormalities

	Male	Female	Total
Translocation	1	2	3
Inversion	1	1	2
Percentage	1.4%	2.11%	3.52%

Table 4: Age

Descriptive Statistics					
N		Minimum	Maximum	Mean	Std. Deviation
Age of female	142	18.0	45.0	30.662	6.3947
Age of male	142	21.0	53.0	35.373	6.2994
Valid N (list wise)	142				

Discussion

In the research, were assessed by Narjes Soltani and co-workers. 43 cases (3.54%), including 25 females and 18 males showed chromosome aberrations. Also, 40 cases, including Robertsonian translocations (4 cases), numerical chromosome aberrations (3 cases) balanced translocations (25 cases), and inversions (10 cases) were found with structural chromosomal abnormality. Polymorphic variants were found in 22 cases [1]. Which was in the northeast of Iran and corresponds with the 3.52% found in 284 patients in Ahwaz.

In study which conducted in Saudi Arabia, 7.6% of cases were carriers of structural or numerical chromosomal abnormalities. Increased frequency of translocations (46%) showed an association with a high RPL incidence. There was a significant relationship between chromosomal abnormalities and patients with consanguineous RPL ($p < 0.05$). Factor V Leiden and Prothrombin A20210G allelic polymorphisms showed a significant association with increased prevalence of RPL [2]. Of 506 cases that were assessed by YALÇIN S, ÖZTÜRK KH, ÖZBAŞ H, and Hilmi O, 15 (2.9%) cases had chromosomal abnormalities. Females were more affected compared to males (respectively 1.9% and 0.98%). Eight out of 15 cases (53.3%) reported structural deviations and two subjects (13.3%) showed numerical abnormalities. Five cases (33.3%) had chromosome variants [3].

In Asoke K. Pal's study Out of 172 couples, 17 ones (9.88%) showed various types of numerical or structural chromosomal abnormalities. Out of 17 couples, eight cases (47.05%) showed balanced translocations, 2 (11.76%) showed Robertsonian translocation, five cases (29.41%) showed the pericentric inversion of chromosomes 8, 9, and Y, and only two women (11.76%) had sex chromosome numerical aberrations. the percentage of chromosomal abnormalities in this study is about 2.8 times more than in our study in Ahvaz. In the study, we detected that 40% of all abnormalities were balanced and 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 found in 121 (3.74%) out of 3235 RPL couples, including 75 females and 46 males at an individual level. 101 subjects had structural aberrations: Robertsonian translocations in 13 (10.7%) subjects, balanced translocations in 46(38.0%) subjects, inversions in 42 (34.7%) subjects and 20(16.5%) subjects had numerical aberrations. 428 non-carriers and 121 carriers were followed up for two years, 229 non-carriers and 55 carriers were subsequently pregnant following diagnosis by intrauterine insemination or natural conception [5].

In the study published in 2018 in Ardabil, Iran 350 patients who had at least two spontaneous abortions were assessed. Eighteen cases (10.28% of couples) had Parental chromosomal abnormalities. 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 results were not significantly different compared to global reports. Thirteen (6.1%) out of 238 couples had chromosomal abnormalities. Twelve chromosomal abnormalities were structural and one case was numerical. In our research, chromosomal abnormalities were observed in 3.52% of couples, which all were structural [7].

In Serapinas. D *et al.* study the rate of chromosomal aberrations in Lithuania affecting RM or the inability to conceive was 3%. The mosaic aberrations and balanced translocations were the most common. Couples unable to conceive mostly had chromosomal aberrations ($p < 0.05$) than other couples (with miscarriages). Overall, 8.1% of patients had miscarriages in the family. No significant difference was found in terms of whether more frequent miscarriages ($p > 0.05$) happened on the female or male side [8].

In the study published in 2006 in Ahvaz by Malekasgar AM and his co-workers, chromosomal assessment showed three abnormal karyotypes (3.8%) in three females and two abnormal karyotypes in conceptions. Two couples were found with consanguineous marriage and the rest of women included one isochromosome for X [46, x, I (xq)], one trisomy '21' (47, xx, +21), two translocations [45, xx, t (15:21)] and [46, xx, t (7:14)], and a ring chromosome (46, xx, r(X)). Also, 27 conceptions were found for these five couples, including 23 abortions with four of them having abortions within the second trimester (21.74%), 18 within the first trimester (78.26%), and three had abnormal children, one had a normal child, and one with stillbirth [9].

In one retrospective study conducted at Balcalı Hospital in Adana, Turkey, karyotype findings of 1510 couples who had recurrent spontaneous abortions were assessed. A total of 62

couples showed chromosome aberrations (4.1%). Chromosome aberrations were detected in 65 cases (24 males and 41 females); structural chromosomal aberrations were detected in 58 cases, which included Robertsonian translocations in 12 subjects, balanced translocations in 30 subjects, inversions in nine cases, deletions in seven subjects, and numerical chromosome aberrations in seven subjects. Structural aberrations, especially translocations, were the commonest kind of aberration in couples with a history of recurrent [10].

In the Franssen MT study that was conducted in the Netherlands Couples with ascertained carrier status following two or more miscarriages showed a decreased risk of viable offspring with unbalanced chromosomal abnormalities. Despite the high risk of miscarriage, their risk of having a normal child is the same as non-carrier couples [11].

In the article published by Elghezal H in Tunisia, 97 chromosome anomalies were found in 1,400 studied couples (6.93%). This prevalence seems to be similar to previous studies in other populations but it is two times more than our study in Ahvaz [12].

Out of 795 couples, 28 couples (3.52%) had a chromosomal abnormality (carrier group). Balanced reciprocal translocations accounted for more than half (65.5%) of the chromosomal abnormalities [13].

In these studies, overall, the probability of abnormal karyotype has been between 3 to 10 percent, which of course depends on the selection of the statical population.

Conclusion

The prevalence of chromosomal abnormalities in our study was 3, 52%, which of course 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

1. Soltani N, Mirzaei F, Ayatollahi H. Cytogenetic Studies of 608 Couples with Recurrent Spontaneous Abortions in Northeastern Iran. *Iranian Journal of Pathology*. 2021;16(4):418.
2. Turki RF, Assidi M, Banni HA, Zahed HA, Karim S, Schulten HJ. Associations of recurrent miscarriages with chromosomal abnormalities, thrombophilia allelic polymorphisms and/or consanguinity in Saudi Arabia. *BMC medical genetics*. 2016;17(1):15-23.
3. Yağın S, Öztürk KH, Özbaş H, Hilmi O. Cytogenetic analysis in couples with recurrent pregnancy loss. *Journal of Surgery and Medicine*. 2021;5(5):449-52.
4. Pal AK, Ambulkar PS, Waghmare JE, Wankhede V, Shende MR, Tarnekar AM. Chromosomal aberrations in couples with pregnancy loss: a retrospective study. *Journal of human reproductive sciences*. 2018;11(3):247.
5. Li S, Chen M, Zheng PS. Analysis of parental abnormal chromosomal karyotype and subsequent live births in Chinese couples with recurrent pregnancy loss. *Scientific Reports*. 2021;11(1):1-10.
6. Jahani Z, Asl H, Akhavan H. Study of Chromosomal Abnormalities in Couples with Recurrent Spontaneous Abortions (RSA) in Ardabil Province. *Journal of*

- Ardabil University of Medical Sciences. 2018;18(1):91-107
7. Houmaid H, El Bekkay C, Nassereddine S, Talbi H, Amehdare L, Hilali A. Chromosomal Abnormalities in 238 Couples with Recurrent Miscarriages in Morocco. *Open Journal of Genetics*. 2018;8:15-22. DOI: 10.4236/ojgen.2018.82002.
 8. Serapinas D, Valantinavičienė E, Machtejevienė E, Bartkevičiūtė A, Bartkevičienė D. Evaluation of chromosomal structural anomalies in fertility disorders. *Medicina*. 2021;57(1):37.
 9. Malekasgar AM, Motlagh ME, Hashemi TM, Ghafari MA. Chromosomal Analysis of Couples with Bad Obstetric Histoty; c2006.
 0. Tunç E, Tanrıverdi N, Demirhan O, Süleymanova D, Çetinel N. Chromosomal analyses of 1510 couples who have experienced recurrent spontaneous abortions. *Reprod Biomed Online*. 2016 Apr;32(4):414-9. DOI: 10.1016/j.rbmo.2016.01.006. Epub 2016 Jan 28. PMID: 26874988.
 10. Franssen MT, Korevaar JC, Van Der Veen F, Leschot NJ, Bossuyt PM, Goddijn M. Reproductive outcome after chromosome analysis in couples with two or more miscarriages: case-control study. *Bmj*. 2006;332(7544):759-63.
 11. Ighezal H, Hidar S, Mougou S, Khairi H, Saâd A. Prevalence of chromosomal abnormalities in couples with recurrent miscarriage. *Fertility and sterility*. 2007;88(3):721-3.
 12. Ynn H, Yan J, Saravelos SH, Li TC. Comparison of reproductive outcome, including the pattern of loss, between couples with chromosomal abnormalities and those with unexplained repeated miscarriages. *Journal of Obstetrics and Gynaecology Research*. 2014;40(1):109-16.