



## **Chromosomal Abnormalities with Recurrent Miscarriage in Couples (A Clinical Study)**

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### **Authors' contributions**

*This work was carried out in collaboration between both authors. Author BJH designed the study, performed the statistical analysis, wrote the protocol and wrote the first draft of the manuscript. Author ZFA managed the analyses of the study. Both authors managed the literature searches.*

### **Article Information**

#### Editor(s):

(1) Dr. B. Eghon Guzman, Hospital Dr. Sotero del Rio, Chile.

#### Reviewers:

(1) Vemula Harshini, Madras Veterinary College, India.

(2) Anupama Mukherjee, Indian Dairy Research Institute, India.

Complete Peer review History: <http://www.sdiarticle4.com/review-history/61557>

**Original Research Article**

**Received 20 July 2020**  
**Accepted 27 September 2020**  
**Published 15 October 2020**

### **ABSTRACT**

The study demonstrated as a cytogenetic study which carried out in Baghdad Governorate on 40 couples have a history of more than 3 recurrent miscarriages, their ages ranged from 20-39 years old for females and 30- 42 years old for males , they came to the ANSEJA medical laboratories in Baghdad for Karyotyping testing . Identifying the shape and structure of the blood couples chromosome using MetaClass Karyotyping system. The result show that : nine cases were found to have abnormal structure of chromosome anomalies while the other 31 cases had a normal Chromosomal complement 5 cases of the tested samples had been show a marker chromosome (47,XX,+mar) , 3 cases of the tested samples show ring chromosome that found in mosaicism (46, XX/47, XX, +r) and the last 2 case was father with Klinefelter syndrome (47,XXY). The study conclude that the chromosomal abnormalities can be possibly evaluated in the patient ,with the history of recurrent spontaneous miscarriage, using conventional cytogenetic detection.

**Keywords:** *Recurrent miscarriage, chromosomal abnormalities.*

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## 1. INTRODUCTION

Miscarriage can be defined as a spontaneous loss of pregnancy more than 3 time in 20 weeks , it is represent the most common complication of pregnancy loss affecting 2– 5% of couples [1]. The high risk of miscarriage can be increases with maternal (age and parity) in other hand the risk of miscarriage increases after one miscarriage, two and three pregnancy losses [2]. Minimum diagnostic workup of couples experiencing consists of a complete history (medical, surgical, genetic and family and a physical examination). The possible origins of recurrent miscarriage are defective policy control of the embryo, uterine structural defaults, immunological dialog between the embryo (or the fetus) and the uterus and with immunological disorders [3]. Many of the studies tried to search variants of genes suspected to be intervening in the different factors of the early maternal fetal loss and the outcome of fertilization, leading to success of implantation and development [4] . Recurrent Miscarriage are considered as an important pathological condition associated with heterogeneous laboratory and clinical findings [5].

Chromosomal abnormalities in couples found to be at risk of repeat miscarriages resulting to lowering the chances to deliver a viable newborn, whoever spontaneous miscarriages that caused by The chromosomal abnormalities may arise from one defective gametes of the parents that will lead to fetal abnormalities and mental disorders [6].

It can be say that 3–6 % of recurrent miscarriage were due to chromosomal abnormalities [7]. pregnancy loss sometimes been the frustrating experience for the tow couples and concern clinician [8]. The most important etiological investigation is Chromosomal analysis of couples which have repeated miscarriages. chromosomal translocations is one of the most important cause of recurrent miscarriage in asymptomatic parents it may result in generation of unbalanced translocation in conspectuses leading to negatively selected by nature and most of the time end with a miscarriage [9]. Minor abnormalities of chromosomal structural are known as polymorphisms which are originated from most of the genetic variations in populations [10]. Nowadays the studies testing many of factors that may increase or decrease the risk of miscarriage, like ; maternal thrombophilic, chromosomal anomalies , anatomic, endocrine, and immunological disorders to identify the causes and preparing for treatment [4]. This study designed to determined the possible etiology of chromosomal anomalies in miscarriage and maternal or paternal genetic abnormality.

## 2. MATERIALS AND METHODS

### 2.1 Samples

The study was carried out in Baghdad Governorate on 40 couples having a history of more than 3 recurrent miscarriages, the ages of these cases was ranged from 20-39 years old for females and 30- 42 years old of males couples with a history of healthy sexual and pregnant properties .



Fig. 1. Karyotyping automatic system used in human cytogenetic studies of chromosomes

## 2.2 Procedure

- a- **Blood collection:** All samples in the presented study were done in the ANSEJA medical laboratories in Baghdad, blood collection made from all patient then by centrifuged (2000 cycle / 10 min) to separate the white blood cell (lymphocyte).
- b- **Karyotyping:** Identifying the shape and structure of the couples chromosome using MetaClass Karyotyping system (Automatic Diagnostic system, made in Barcelona, Spain (Fig. 1)) it is a completely automatic system that enables us to classify chromosomes from metaphase preparations with aims of special media Lymphoprime (complete medium for cultivation of peripheral blood

Lymphocytes ) (Capricorn Scientific GmbH , German).

## 3. RESULTS

According to G-banding analyses of 40 cases of study, nine cases were found to have chromosomal abnormalities , while the other 31 cases had a normal Chromosomal complement (Table 1).

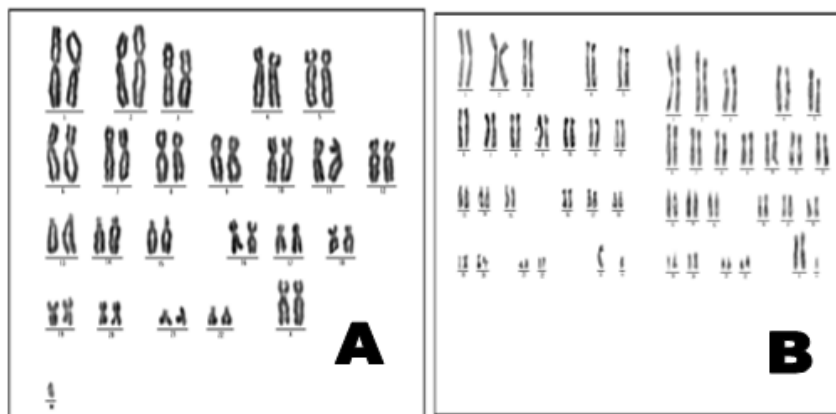
The result of chromosomal abnormalities can be demonstrated as the following : 5cases of the tested samples showed a marker chromosome (47,XX,+mar) (Fig. 2.a) , (3) cases of the tested samples show ring chromosome that found in mosaicism (46, XX/47, XX, +r) (Fig. 2.b) and in the last ( 2 ) cases, father were found to have Klinefelter syndrome (47,XXY) (Fig. 2.c) (Table 2) .

**Table 1. Numbers and percentage of normal and abnormal karyotype according to G-band Analyses**

No	Number of cases	Percentage %
No. of normal chromosome	31	77.5%
No. of abnormal chromosome	9	22,5%
Total :	40	100%

**Table 2. Demonstrate the Karyotype of chromosome and age of each case at once**

Case no.	Gender	Karyotypes	Age	No. of Miscarriages
1	Female	47, XX,+mar	33	3
2	Female	47, XX,+mar	40	3
3	Female	47, XX,+mar	30	2
4	male	47, XY,+mar	29	2
5	male	47, XY,+mar	30	2
6	Female	46, XX/47, XX, +r	28	5
7	Female	46, XX/47, XX, +r	36	4
8	Male	46, XY/47, XX, +r	34	4
9	Male	47, XXY	35	3



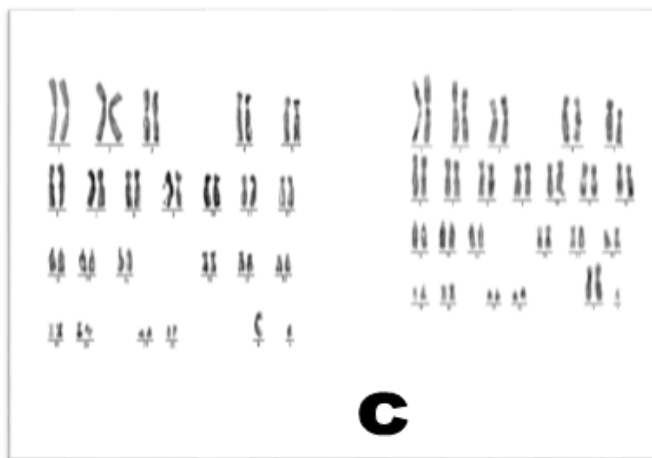


Fig. 2. (A,B,C). Show the types and figure of the 3 abnormal chromosomal distribution which cause of recurrent Miscarriage

#### 4. DISCUSSION

As what we said before the chromosomal abnormalities can be demonstrated firstly as the (a marker chromosome (47,XX,+mar) (Fig. 2.a) , (3) cases of the tested samples show ring chromosome that found in mosaicism (46, XX/47, XX, +r) (Fig. 2.b) and in the last ( 2 ) cases, father were found to have Klinefelter syndrome (47,XXY ) which are some of chromosomal anomalies could be cause recurrent miscarriage , the result was agreed with Atia [11] when he concluded that genetic profile of miscarriages is important for prognosis and potential counseling planning, as well as the prenatal diagnostic strategy in subsequent pregnancies. And Aayed et al. [12] when they reported that chromosomal abnormality is one of the important factors contributing to recurrent miscarriage, they found that we found 4/14 cases with X chromosome mosaicism. Monosomy X cell's rate varied between 6% and 22%. . the result was not agreed with Kolben et al. [13], they reported that PPAR depletion leads to fetal loss in early pregnancy due to the missing PPAR expression and extended placental defects

#### 5. CONCLUSION

The study conclude that the chromosomal abnormalities can be possibly evaluated in the patient with the history of recurrent spontaneous miscarriage using conventional cytogenetic detection.

#### COMPETING INTERESTS

Authors have declared that no competing interests exist.

#### REFERENCES

1. Dean Deepika Delsa Sarita Agarwal and Poonam Tripathi. Connecting links between genetic factors defining ovarian reserve and recurrent miscarriages, J. Assis. Reprod. and Genetics. 2018;(35):2121–2128.
2. Vidyadhari M, Sujatha M, Krupa P, Nallari Pratibha, Venkateshwari A. Association of genetic polymorphism of vascular endothelial growth factor in the etiology of recurrent pregnancy loss: a triad study. J. Ass. Reprod. and Gen. 2019;36:979–988.
3. Vaiman Daniel. Genetic Regulation of Recurrent Spontaneous Abortion in Humans, Biomed J. 2014;38(1):11-27.
4. Torres MH, Gimenez C. Garrido, Reig Alijotas. Genetics of recurrent miscarriage and fetal loss. Best Practice & Research Clinical Obstetrics and Gynaecology. 2017;42:11-25.
5. Kling Christiane, Jürgen Hedderic and Dieter Kabelitz. Fertility after recurrent miscarriages: results of an observational cohort study. Arch Gynecol Obstet. 2018;297:205–219.
6. Rull Kristiina, Liina Nagirnajaand MarisLaan. Genetics of recurrent miscarriage: Challenges, current

- knowledge ,future directions , Epigenomics. 2012;3(34):1-13.
7. Turki Rola F, Mourad Assidi2, Huda A. Banni , Hanan A. Zahed, Sajjad Karim, Hans-Juergen Schulten, Muhammad Abu-Elmagd, Abdulrahim A. Rouzi1, Osama Bajouh , Hassan S. Jamal, Mohammed H. Al-Qahtani and Adel M. Abuzenadah. Associations of recurrent miscarriages with chromosomal abnormalities, thrombophilia allelic polymorphisms and/or consanguinity in Saudi Arabia. J. BMC Medical Genetics. 2016;17(1):69.
  8. De-Marqui Alessandra Bernadete Trovó. Chromosomal abnormalities in recurrent miscarriages by conventional karyotyping analysis . J. Rev. Bras. Saúde Mater. Infant., Recife. 2018;18(2): 265-276
  9. Kacprzak Marta, Marta Chrzanowska, Beata Skoczylas, Hanna Moczulska, Maciej Borowiec and Piotr Sieroszewski1. Genetic causes of recurrent miscarriages, J. Ginekologia Polska. 2016;87(10):722–726.
  10. Ocak Z, Özlü T, Ozyurt. Association of recurrent pregnancy loss with chromosomal abnormalities and hereditary thrombophilias. J. African Health Sciences. 2013;13(2):447-452.
  11. Atia A Tarek. Overview of genetic causes of recurrent miscarriage and the diagnostic approach. J. BIOCELL. 2019;43(4): 253-262
  12. Ayed Wiem, Islem Messaoudi1, Zouhour Belghith, Wajih Hammami1, Imen Chemkhi1, Nabila Abidli1, Helmy Guermani1, Rim Obayl and Ahlem Amouri. Chromosomal abnormalities in 163 Tunisian couples with recurrent miscarriages , Pan African Medical Journal. 2017;28:99. DOI: 10.11604/pamj.2017.28.99.11879
  13. Kolben Theresa Maria, Elisabeth Rogatsch, Aurelia Vattai, Anna Hester, Christina Kuhn, Elisa Schmoeckel, Sven Mahner, Udo Jeschke and Thomas Kolben. PPAR Expression Is Diminished in Macrophages of Recurrent Miscarriage Placentas . international of molecular science. 2018;19:1872. DOI: 10.3390/ijms19071872

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