

Chromosomal Aberrations in Couples with a history of Recurrent pregnancy Loss: The status in Iran

Presented by:

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Outline

- **Introduction**
- **Aim of the study**
- **Design**
- **Key findings**
- **Discussion**

Introduction

- RPL defined as two or more consecutive loss of pregnancies before 20–22 weeks of gestation
- Determining the etiology of RPL can be an important step in managing future pregnancies and subsequent interventions
- The exact etiology of the human reproduction problems have not been definitely clarified
- several underlying factors including genetic makeup, uterine abnormalities, hormonal imbalances, immunological disorders, and environmental factors

Aim & Study Design

The frequency of chromosomal abnormalities in couples with a history of RPL varies significantly among different populations → we reviewed the frequency of chromosomal anomalies in individuals suffering from RPL in Iran

Study design:

- Keywords: Recurrent pregnancy loss/abortion [AND] chromosome abnormality/cytogenetic analysis [AND] Iran
 - Pubmed.gov, Scopus.org and google scholar
- applying filters
- Extract the data
- Review the major findings

A cytogenetic study of couples with repeated spontaneous abortions

Shirin Niroumanesh, Parvin Mehdipour, Ali Farajpour, Soodabeh Darvish

- **100 couples with recurrent abortions**
- **Eight females (8%) & five males (5%) have abnormal karyotypes**
 - **69.2% abnormal karyotypes**
 - **30.8%: chromosomal variants**
- **(30.8%) balanced reciprocal translocations, (23%) Robertsonian translocations, (23%) pericentric inversions, (7.7%) paracentric inversion, (7.7%) chromosomal marker**

Chromosomal Analysis of Couples with Repeated Spontaneous Abortions in Northeastern Iran

Saeedeh Ghazaey, M.Sc.^{1,2}, Fatemeh Keify, M.Sc.², Farzaneh Mirzaei, M.Sc.², Masumeh Maleki, M.Sc.², Semiramis Tootian, B.Sc.², Mitra Ahadian, B.Sc.², Mohammad Reza Abbaszadegan, Ph.D.^{2,3*}

- **six-year period from 2005 to 2011**
- **728 couples**
- **11.7% of couples were carriers of chromosomal aberrations**
- **Balanced reciprocal translocations were the most frequent chromosomal anomalies (62.7%) detected in current study**

ORIGINAL ARTICLE

Evaluation of 1100 couples with recurrent pregnancy loss using conventional cytogenetic, PGD, and PGS: hype or hope

Kamelia Farahmand^{1,2*}, Hamid Kalantari^{2*}, Mostafa Fakhri², Abolhasan Shahzadeh Fazeli^{2,3}, Shabnam Zari Moradi², Navid Almadani², Mehrdad Hashemi¹, Hamid Gourabi², and Anahita Mohseni-Meybodi²

- 2008 and 2014
- 1100 couples
- The frequency of chromosomal abnormalities in these patients was 4.95%
- Women demonstrated more abnormalities (6.82%) in comparison to men (3.09%).

Original Article

Hum Reprod Sci 2020;13:216-20.

Cytogenetic Analysis of 570 Couples with Recurrent Pregnancy Loss: Reporting 11 Years of Experience

Reza Alibakhshi, Parham Nejati¹, Sara Hamani², Narges Mir-Ahadi², Nazanin Jalilian

- 570 couples with two or more spontaneous abortions
- 2008 to 2018
- 11.5% had chromosomal aberrations
- chromosomal aberrations were found in 62 (11%) of females and 67 (11.8%) of males

Table 3: Classification of chromosome abnormalities among all referred patients with recurrent pregnancy loss

Karyotype	Type of chromosomal aberration	Total number of cases	Male/female
Normal karyotype		1011	503/508
Chromosomal aberrations		129	67/62
	Numerical abnormality	1	1/0
	Structural abnormality	18	6/12
	Chromosome polymorphisms	110	61/59

Table 5: Distribution of chromosome polymorphisms in recurrent miscarriages cases

Variant	Number of cases (male:female)
1qh+	26 (12:14)
9 qh+	14 (6:8)
9 qh-	2 (1:1)
16 qh+	5 (4:1)
13ps+	7 (3:4)
14 ps+	3 (2:1)
14 pstk+	5 (1:4)
15 ps+	6 (2:4)
21 ps+	9 (3:6)
21pstk+	1 (1:0)
22ps+	6 (5:1)
22pstk+	2 (0:2)
Yqh+	9
Yqh-	12
Inv(9)	3 (0:3)
Total	110



Association Study of Recurrent Abortion With Chromosomal Abnormalities and Mutation of Prothrombin Gene in 100 Affected Women in the Northwest of Iran

Seyed Ali Rahmani^{1*}, Reihaneh Amiri², Parisa Mosapour³

- 100 couples
- 2015-2017
- 5% were diagnosed with chromosomal abnormalities
 - 1 Robertsonian translocation (45, XX, rob(14;21))

Cytogenetic Studies of 608 Couples with Recurrent Spontaneous Abortions in Northeastern Iran

Narjes Soltani¹, Farzaneh Mirzaei², Hossein Ayatollahi^{1,2*}

- **608 couples**
- **2010-2019**
- **~6% were carriers of chromosomal aberrations**
- **balanced translocations**

Risk Factors Associated with Recurrent Pregnancy Loss and Outcome of Pre-Implantation Genetic Screening of Affected Couples

Nayeralsadat Fatemi, Ph.D.^{1, 2#}, Maryam Varkiani, M.Sc.^{2, 3#}, Fariba Ramezanali, M.D.⁴, Babak Babaabasi, M.Sc.², Azadeh Ghaheri, Ph.D.⁵, Alireza Biglari, Ph.D.^{1*}, Mehdi Totonchi, Ph.D.^{2, 6*}

- **602 Iranian couples**
- **15.61% chromosomal abnormalities in RPL couples**
- **8.13% chromosomal abnormalities in RPL patients**
- **reciprocal translocations were more frequent (2.74%)**

Chromosomal Study of Couples with the History of Recurrent Spontaneous Abortions with Diagnosed Blighted Ovum

Sahar Shekoochi, Majid Mojarrad, Reza Raoofian, Shahab Ahmadzadeh, Salmah Mirzaie, Mohammad Hassanzadeh-Nazarabadi*

- 68 couples with the history of spontaneous abortion (diagnosed blighted ovum)
- 2007-2012
- Increased consanguineous marriages in blighted ovum suffering couples higher (68.5% versus 31.5% ,*P value* <0.001)
- balanced chromosomal rearrangements:
 - 8.3 in non-consanguineous couples
 - 2.3% of RSA affected couples with consanguineous marriage

Study	Year	Region	Number of participants	Chromosomal aberrations (%)	structural chromosomal rearrangements (%)	normal variants (%)
Niroumanesh et al	2011	Mirza Kouchak Khan hospital, Tehran	100 couples	6%	69.2%	30.8%
Ghazaey et al	2005-2011	North eastern Iran	728 couples	11.7 %	58%	42%
Farahmand et al.	2008-2014	Royan Reproduct ive Center	1100 couples	8%	62%%	38%%
Rahmani, et al.	2015-2017	Northwest Iran	100 couples	5%	20%	80%
Alibakhshi , et al.	2008-2018	Kermansh ah	570 couples	11.5%	15%	85%
Fatemi et al	2006- 2018	Royan Reproduct ive Clinic	602 couples	15.61	52%	48%
Soltani N, et al.	2010-2019	Mashahd	608	5.54%	64%	36%

Discussion

- **Varying frequencies of chromosomal aberrations**
 - ✓ **5-15%**
 - ✓ **Different Normal/abnormal classification rules**
 - ✓ **Size of the study population**
 - ✓ **Technique resolution**
 - ✓ **Ethnicity??**
 - ✓ **Consanguinity**

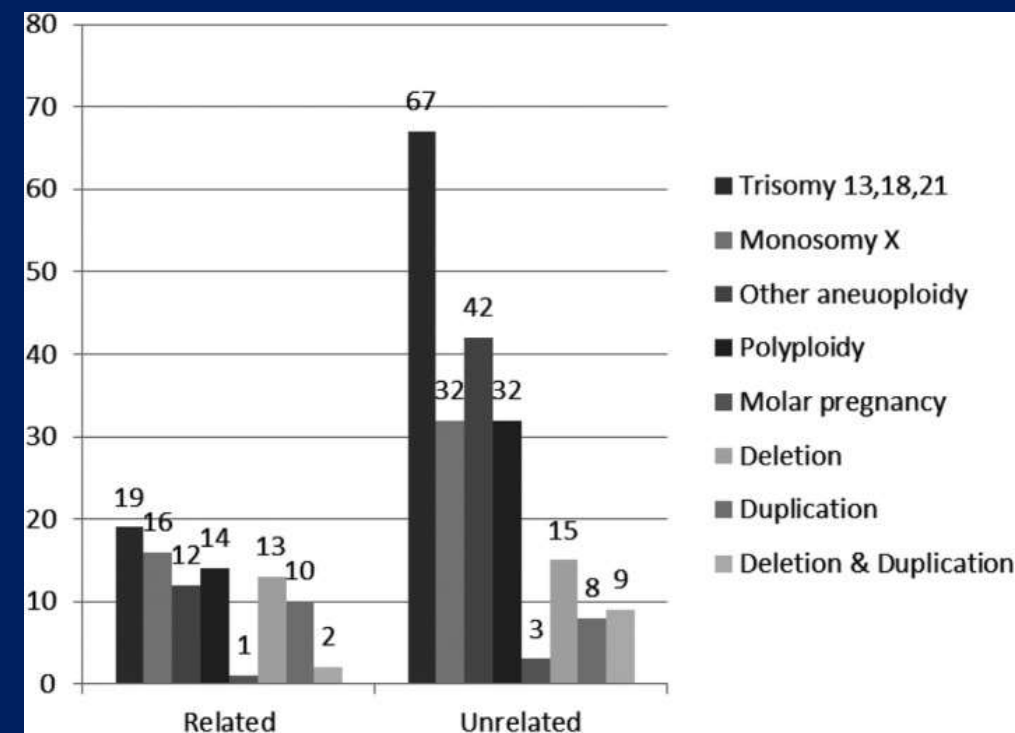
Discussion

- **Varying frequencies of structural aberrations**
 - ✓ **15-69.2%**
 - ✓ **Different Normal/abnormal classification rules**
 - ✓ **Size of the study population**
 - ✓ **Technique resolution**
 - ✓ **Reporting policy**
 - ✓ **Genetic background**

Chromosomal aberrations in pregnancy and fetal loss: Insight on the effect of consanguinity, review of 1625 cases

Kimia Najafi^{1,2}  | Soheila Gholami¹ | Azadeh Moshtagh¹ | Masood Bazrgar¹  |
Neda Sadatian¹ | Golemaryam Abbasi¹ | Parvin Rostami³ | Soheila Khalili³ |
Mojgan Babanejad^{2,3} | Bahareh Nourmohammadi³ | Negin Faramarzi Garous³ |
Hossein Najmabadi^{2,3} | Roxana Kariminejad¹ 

- 1625 products of abortion or fetal tissue
- Positive correlation of consanguinity and recurrent abortion (Correlation Coefficient 0.294, Sig 0.000)
- a significant difference in frequency of imbalances in related versus unrelated couples



Discussion

- **Most frequent Aberration:**
 - **Reciprocal translocation**
 - **Farahmand et al:**
 - **numerical abnormalities (59.63%)**
- **Most frequent heteromorphism:**
 - **Not consistent**
 - **1qh+ in Kermanshah vs inv(9) in Northeastern Iran**

Conclusion

- **Varying frequencies of chromosomal aberrations**
 - **Both abnormalities & normal variants**
 - **Possible effect of genetic background?**
- **Balanced reciprocal translocation: the most frequent aberration**
 - **Except for 1 study**

Suggestion

- Individual frequencies in different ethnic groups
- A comprehensive analysis on total frequencies in Iranian population
- Revisiting normal variants
- The importance of consanguinity!!

Thank you

