

Letter to Editor

Asian Pacific Journal of Reproduction

Journal homepage: <https://journals.lww.com/apjr/>



doi: 10.4103/apjr.apjr_17_26

Rare chromosomal rearrangements among couples with recurrent pregnancy loss: An observational study at a tertiary care centre

Saji K.G.¹, Soumya Raj¹, Bindu Menon², Ragitha T.S.¹, Sareena Gilvaz², Suresh Kumar Raveendran^{1✉}

¹Jubilee Centre for Medical Research, Jubilee Mission Medical College & Research Institute, Thrissur, Kerala, India

²Department of Obstetrics and Gynaecology, Jubilee Mission Medical College & Research Institute, Thrissur, Kerala, India

Recurrent pregnancy loss (RPL) affects 2%-5% of couples attempting to conceive. It is a highly heterogeneous condition attributed to several factors including endocrine dysfunction, autoimmune disorders, thrombophilia, genetic abnormalities, infectious diseases, uterine anomalies, sperm DNA fragmentation, and epigenetics. Among genetic causes, chromosomal abnormalities are the most frequent etiological factor of early miscarriage, accounting for 50%–60% of first trimester abortions. Numerical or structural chromosomal changes may result in spontaneous miscarriages. These anomalies arise as a result of chromosomal translocation, non-disjunction, or mutations[1]. Transmission of parental chromosomal abnormalities may be one of the chances for a recurrence of miscarriage in the first trimester of pregnancy, albeit the cause is unknown[2,3].

Balanced rearrangements do not gain or lose genetic material. However, it causes unbalanced karyotypes in gametes during meiosis, resulting in recurrent abortions. These abnormalities result in spontaneous abortions because they are fatal to the growing embryo. Parental karyotyping is helpful to ascertain whether a parent has such a rearrangement since it explains the losses and offers information about the possibility of future miscarriages.

The present study aims to assess the frequency and types of chromosomal abnormalities in couples with RPL in a South Indian Tertiary Care Centre and to evaluate the role of cytogenetic analysis in elucidating the underlying causes of recurrent miscarriages. The study was conducted between September 2017 and August 2025, in couples with recurrent abortions who were referred from our Obstetrics and Gynaecology department and approved by the local institutional ethics committee.

A total of 188 individuals (94 couples) were investigated for chromosomal abnormalities, who had two or more consecutive pregnancy losses before 20 weeks of gestation. Informed consent was obtained from the couples before performing the investigation. Metaphase chromosome preparations from the peripheral blood cultures were made according to the standard cytogenetic protocols. Chromosome analysis was performed by GTG-Banding (G-bands

by Trypsin using Giemsa) at approximately 450-band level. Thirty metaphases were scanned for each individual, but for suspicious cases (mosaicism, etc.) extended up to fifty. Karyotypes were described according to the International System for Cytogenetic Nomenclature (ISCN, 2024).

In the current study, the age of the couples ranged from 20 to 58 years, with a mean maternal age of 31.0 years and paternal age of 29.9 years. The number of miscarriages ranged from two to six. Among 94 couples, abnormal karyotypes were detected in 14 couples (14.9%), including 8 women (57.1%) and 6 men (42.9%).

Out of 14 chromosomal abnormalities, 6(42.9%) (Table 1) were reciprocal translocations and remainings were known polymorphic variants. Reciprocal translocations were observed in 6 cases included 4 (66.7%) in women and 2 (33.3%) in men. Among the 6 reciprocal translocations, 4 of them were novel translocation such as t(1;16)(q21;q12.1), t(6;11)(p25;q23), t(21,22)(q21;q12), and t(15;17)(q26.1;q25) (Figure 1). Polymorphic variants observed in the study group were inversion in chromosome 9 and Y, heterochromatin variant in 9th chromosome, satellite in chromosomes such as chromosome #15, 21 and 22. Studies have revealed that chromosomal heteromorphism is more common in RPL as well as couples who are infertile[4]. Pericentric inversion was also associated with RPL. During crossing over, pericentric inversion may result in deletion or duplication of a segment in the involved chromosome[5].

✉To whom correspondence may be addressed. E-mail: drsureshkumar@jmmc.ac.in

This is an open access article distributed under the terms of the Creative Commons Attribution-Non-Commercial-No Derivatives License 4.0 (CCBY-NC-ND), where it is permissible to download and share the work provided it is properly cited. The work cannot be changed in any way or used commercially without permission from the journal.

For reprints contact: reprints@medknow.com

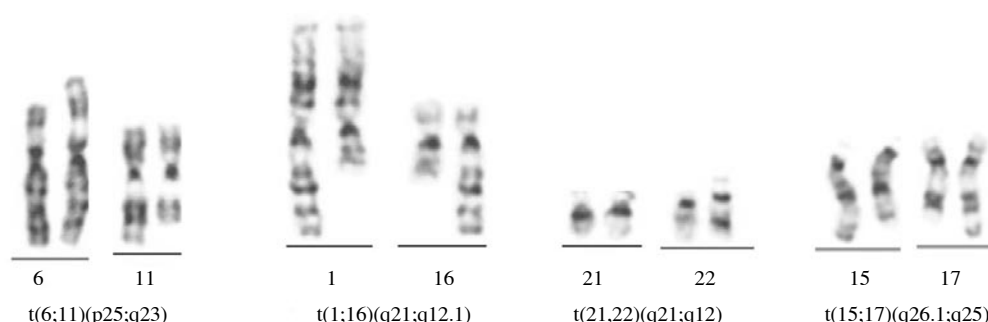
©2026 Asian Pacific Journal of Reproduction Produced by Wolters Kluwer- Medknow.

How to cite this article: Saji KG, Raj S, Menon B, Ragitha TS, Gilvaz S, Raveendran SK. Rare chromosomal rearrangements among couples with recurrent pregnancy loss: An observational study at a tertiary care centre. *Asian Pac J Reprod* 2026; 15(2): 97-98.

Article history: Received: 17 January 2026; Revision: 11 March 2026; Accepted: 16 March 2026; Available online: 14 April 2026

Table 1. Cytogenetic findings in couples with miscarriages.

Couple No.	Number of miscarriages	Age of female (years)	Female karyotype	Age of male (years)	Male karyotype
1	2	21	46,XX,t(2;3)(q33;q27)	30	46,XY
2	2	32	46,XX,t(2;19)(p15;q13.1)	37	46,XY
3	6	30	46,XX,t(1;16)(q21;q12.1)	40	46,XY
4	2	23	46,XX	30	46,XY,t(6;11)(p25;q23)
5	2	22	46,XX	33	46,XY,t(21;22)(q21;q12)
6	3	30	46,XX,t(15;17)(q26.1;q25)	32	46,XY

**Figure 1.** Novel translocations observed in recurrent pregnancy loss cases.

The type of rearrangement among the chromosomes determines the chance of a subsequent healthy live birth. Cytogenetic investigations detect genomic constitution of an individual and rely on this to identify genetic defects. When any of partner has such structural anomaly, prenatal diagnosis helps to identify the genetic make-up of offspring. Preimplantation genetic diagnosis (PGD) may also be beneficial for the couples for the transfer of unaffected embryo or healthy donor gametes.

Conflict of interest statement

The authors declared that there is no conflict of interest in this study.

Acknowledgement

We acknowledge the Jubilee Centre for Medical Research (JCMR) for providing all the study facilities.

Funding

The study received no extramural funding.

Authors' contributions

Saji K.G. and Suresh Kumar Raveendran made the conceptualization; Soumya Raj and Ragitha T.S. conducted the methodology; Bindu Menon and Sareena Gilvaz were responsible for data collection; Soumya Raj, Ragitha T.S., and Suresh Kumar

Raveendran carried out data analysis; Saji K.G. contributed to writing original draft version; Soumya Raj and Suresh Kumar Raveendran performed supervision; all the authors were responsible for review and editing.

Publisher's Note

The Publisher of the *Journal* remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

References

- [1] Gonçalves RO, Santos WV, Sarno M, Cerqueira BA, Gonçalves MS, Costa OL. Chromosomal abnormalities in couples with recurrent first trimester abortions. *Rev Bras Ginecol Obstet* 2014; **36**(3): 113-117.
- [2] Christiansen OB, Nybo Andersen AM, Bosch E, Daya S, Delves PJ, Hviid TV, et al. Evidence-based investigations and treatments of recurrent pregnancy loss. *Fertil Steril* 2005; **83**: 821-839.
- [3] Flynn H, Yan J, Saravolos SH, Li TC. Comparison of reproductive outcome, including the pattern of loss, between couples with chromosomal abnormalities and those with unexplained repeated miscarriages. *J Obstet Gynaecol Res* 2014; **40**: 109-116.
- [4] Hong Y, Zhou YW, Tao J, Wang SX, Zhao XM. Do polymorphic variants of chromosomes affect the outcome of *in vitro* fertilization and embryo transfer treatment? *Hum Reprod (Oxford, England)* 2011; **26**: 933-940.
- [5] Fauth C, Bartels I, Haaf T, Speicher MR. Additional dark G-band in the p-arm of chromosome 19 due to a paracentric inversion with a breakpoint in the pericentromeric heterochromatin. *Am J Med Genet* 2001; **103**(2): 160-162.