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ORIGINAL ARTICLE

A Prospective Descriptive Study of Chromosomal Abnormalities in First Trimester Abortions

Rudragouda S Bulagouda, Neelamma Patil*, Darshana N Shah, Gurushantappa S Kadakol*****

Abstract

Introduction: Chromosomal abnormalities, particularly aneuploidies, are the leading cause of first-trimester pregnancy loss, a common reproductive problem. Understanding these genetic factors is essential for counseling and management of affected couples. **Objectives:** To analyze chromosomal profiles of first-trimester abortion cases and identify the most prevalent abnormalities causing miscarriage. **Materials & Methods:** A prospective cytogenetic study was conducted on 122 first-trimester abortion cases. Peripheral blood samples were processed for conventional karyotyping with Giemsa banding. Chromosomal anomalies were examined using a trinocular fluorescent microscope with Gene Asis software (Olympus BX53). Maternal age was recorded and correlated with outcomes. **Results:** Among 122 cases, 35.2% (n=43) showed abnormalities, while 64.7% (n=79) had normal karyotypes. Trisomy was most frequent (60.4%), with Trisomy 21 (42.3%), Trisomy 13 (34.6%), and Trisomy 16 (23.7%) predominating. Polyploidy (23.2%), Monosomy X (11.6%), and structural abnormalities (9.3%) were also detected. Abnormalities were more common in women ≥ 30 years, especially trisomies, whereas Monosomy X and polyploidy occurred more in younger women. **Conclusion:** Chromosomal abnormalities, particularly autosomal trisomies, are major causes of first-trimester pregnancy loss and strongly correlate with maternal age. Incorporating cytogenetic testing into miscarriage evaluation, especially in recurrent cases, can aid reproductive planning. Advanced genomic tools may enhance diagnostic accuracy.

Key Words

First-trimester Abortion, Chromosomal Abnormalities, Trisomy, Monosomy X, Polyploidy, Maternal Age, Cytogenetics.

Introduction

Globally, around 23 million pregnancy losses occur each year, representing 10–15% of clinically recognized pregnancies[1]. Most losses happen before 8–9 weeks of gestation, and many additional early losses remain undetected. Overall, 10.8% of women experience at least one miscarriage, while 1.9% and 0.7% suffer two or three, respectively[2]. Identifying the cause of pregnancy loss is vital for guiding management, prognosis, and counseling for future pregnancies.

The majority of first-trimester losses are attributed to genetic abnormalities of the embryo, particularly aneuploidy, which results from an abnormal number of chromosomes[3]. Aneuploidies commonly arise during oogenesis and in the early embryonic cell divisions[4]. The risk increases with advancing maternal age due to meiotic segregation errors and DNA replication stress

Department of Anatomy, *Department of Obstetrics and Gynaecology, **Research Scholar, ***Genetics Laboratory, BLDE (DU) SBMPMC Vijayapura, Karnataka, India.

Correspondence to: Dr Gurushantappa S Kadakol, Assistant Professor, School of Applied Science and Technology, Genetics Laboratory, Dept. of Anatomy, BLDE (DU) SBMPMC Vijayapura, Karnataka, India.

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during early cleavage divisions. Chromosomal instability (CIN), although common in embryos, is absent at birth, suggesting that post-zygotic chromosomal errors and meiotic aneuploidies often lead to implantation failure or early miscarriage[5]. Embryos with stable genomes are more likely to survive.

First-trimester miscarriages, though clinically detectable, are frequent and often underreported. Studies show that chromosomal abnormalities account for up to 70% of these losses[6,7]. Spontaneous miscarriages occur in 15–20% of pregnancies, with a significant proportion due to fetal chromosomal defects. Parental chromosomal abnormalities also contribute to recurrent pregnancy loss; 2–8% of couples experiencing repeated miscarriages harbor structural rearrangements[8,9].

The causes of miscarriage are multifactorial and include congenital malformations, infections, endocrine dysfunction, autoimmune conditions, environmental exposures, and chromosomal defects. In couples with recurrent loss, balanced chromosomal rearrangements such as translocations and inversions are frequently implicated[10]. These can generate gametes with unbalanced chromosomal content due to abnormal crossing-over during meiosis. Such imbalances typically result in embryonic lethality, spontaneous abortion, or early neonatal death[10].

Our study aimed to investigate the cytology and etiology of abortion cases in this area. The current study specifically sought to determine the most prevalent chromosomal defects causing these pregnancy losses and to assess the chromosomal structure in first-trimester abortion cases.

Objectives

Primary Objective

To analyze the chromosomal profiles of first-trimester abortion cases.

Secondary Objective

To determine the most prevalent chromosomal abnormalities and their association with maternal age, in order to provide cytogenetic insights useful for counseling and reproductive planning.

Methodology

Study Design and Setting

This was a **prospective cytogenetic observational study** conducted in the Department of Obstetrics and Gynaecology in collaboration with the Genetics Laboratory, Department of Anatomy, BLDE (Deemed to be University), Shri B. M. Patil Medical College, Hospital and Research Centre, Vijayapura, Karnataka, India. The study was carried out between [insert study duration, e.g., January 2018 to December 2023].

Ethical Approval and Consent

The study protocol was reviewed and approved by the Institutional Ethical Committee of BLDE (DU) (Approval No. BLDE(DU)/IEC-SBMPMC/224/2017-18 dated 21/09/2017). Written informed consent was obtained from all participants prior to inclusion in the study.

Study Population and Sampling Technique

The study included women presenting with first-trimester pregnancy loss. A **consecutive sampling technique** was applied, whereby all eligible women who met the inclusion criteria were enrolled until the required sample size (n = 122) was achieved.

Inclusion Criteria

1. Gestational age <14 weeks.
2. Missed abortions.
3. Blighted ovum.
4. Spontaneous abortions.
5. Medical termination of pregnancy for abnormal fetus.
6. Willingness to provide informed consent.

Exclusion Criteria

1. Gestational age >14 weeks.
2. Medical termination of pregnancy for an otherwise normal fetus.
3. Patients unwilling to participate.

Sample Size Calculation

The sample size of 122 was calculated based on the estimated prevalence of chromosomal abnormalities in recurrent miscarriages (5%) at a 99% confidence level and 5% absolute error.

Sample Collection and Processing

Peripheral blood (1 ml) was collected from each participant into sterile sodium heparinized vacutainers. Samples were transported immediately to the Genetics Laboratory for processing.

Cell Culture and Harvesting

Cell culture was initiated within 24 hours of collection using 9 ml RPMI complete medium, 100 µl phytohemagglutinin (PHA), 10 µl penicillin-streptomycin, and 1 ml blood sample in 50 ml culture flasks. Cultures were incubated for 72 hours at 37°C in a CO_2 incubator. After incubation, cells were harvested using hypotonic KCl treatment followed by fixation with Carnoy's fixative.

Slide Preparation and Staining

Cell suspensions were dropped onto pre-cleaned glass slides from a height of ~3 feet to spread chromosomes. Air-dried slides were treated with trypsin-EDTA for 2 seconds, rinsed, and stained with Giemsa for 10 minutes.

Karyotyping and Analysis

Slides were examined under a trinocular fluorescent microscope (Olympus BX53) equipped with Gene Asis

software. At least 20 metaphases per sample were analyzed for numerical and structural abnormalities. Chromosomal findings were recorded as per the International System for Human Cytogenomic Nomenclature (ISCN).

Statistical Analysis

Data were entered in Microsoft Excel and analyzed using SPSS software (version XX). Frequencies and percentages were calculated for different chromosomal abnormalities. Mean maternal age (±SEM) was compared between normal and abnormal karyotype groups, and stratified analysis was performed across maternal age categories.

In this study, the term *abnormal abortion* refers to first-trimester pregnancy losses with chromosomal abnormalities identified on karyotyping, while *normal abortion* refers to cases with normal karyotype results.

Results

Cytogenetic analysis of 122 first-trimester abortion cases revealed that **64.7% (n=79)** had a normal karyotype, while **35.2% (n=43)** exhibited chromosomal abnormalities. The average mother's age for typical abortions was **28.11 ± 0.64 years**, compared to **27.00 ± 0.78 years** for abnormal cases (Table 1) was calculated by Standard Error Mean (SEM).

Among the 43 abnormal cases, **trisomy** was the most prevalent **(60.4%, n=26)**, followed by **polyploidy (23.2%, n=8), Monosomy X (11.6%, n=5)**, and **structural anomalies (9.3%, n=4)** (Table 1)

Trisomy 21 was the most frequent **(42.3%, n=11)**, followed by Trisomy 13 **(34.61%, n=9)** and Trisomy 16 **(23.7%, n=6)**. The highest average mothers age for Trisomy 21 **(28.30 ± 1.80 years)** (Table 2).

The **25–29-year age group** had the highest number of abnormal abortions **(n=19, 15.5%)**, with trisomy predominating **(n=9, 7.3%)**. The **30–34-year group** showed a marked increase in trisomy **(n=12, 9.83%)**, while Monosomy X was more common in younger women **(19–24 years)** (Table 2)

The study revealed a strong association correlation between **trisomy prevalence and advanced maternal age**, with a notable increase in cases among women aged **30 years and above.** In contrast**, Monosomy X and polyploidy** were more frequently observed in younger women **(19–29 years)**, suggesting distinct etiological mechanisms for these abnormalities compared to age-related trisomies. Additionally, **structural anomalies**, though rare, were associated with older maternal age, as evidenced by a mean age of **30.50 ± 2.37 years** among affected cases. These findings underscore the substantial influence of maternal age on the kind and frequency of chromosomal abnormalities in abortions performed during the first trimester.

Discussion

The present research elucidates the significant role of chromosomal abnormalities' part in the genesis of first-trimester spontaneous abortions. Cytogenetic analysis revealed a 35.2% prevalence of chromosomal aberrations, corroborating earlier reports which estimate

Table 1 : Distribution of normal and chromosomally abnormal abortions with Mean Maternal Age.

Category	n%	Mean Maternal Age (±SEM)
Normal karyotype Abortions	79 (64.7%)	28.11 ± 0.64
Chromosomally Abnormal Abortions	43 (35.2)	27.00± 0.78
Trisomy (all types)	26 (60.4 abnormal)	30.12 ± 1.10
-Trisomy 21	11 (42.3 trisomy)	28.30 ± 1.80
-Trisomy 13	09 (34.6 trisomy)	29.13 ± 1.92
-Trisomy 16	06 (23.1 trisomy)	28.33 ± 2.04
Polyploidy (2n/3n/4n)	08 (23.2 abnormal)	24.0-30.7 (range)
Monosomy	05 (11.6 abnormal)	23.60 ± 0.68
Structural abnormalities	04 (9.3 abnormal)	30.50 ± 2.37
Total Cases	**122 (100)**	

Table 2 : Chromosomal Abnormalities in First-Trimester Abortions Stratified by Maternal Age Group.

Maternal age group (years)	Total abortions (n)	Abnormal abortion (n,%)	Trisomy (n)	Monosomy X (n)	Polyploidy (n)	Structural (n)
19-24	36	01(0.8)	-	01	-	-
25-29	42	19(15.5)	09	02	03	05
30-34	28	17(13.9)	12	03	-	02
35-39	09	05(4.1)	09	01	-	01
≥40	07	01(0.8)	-	-	-	01
Total	**122**	**43 (35.2)**	**24**	**07**	**03**	**09**

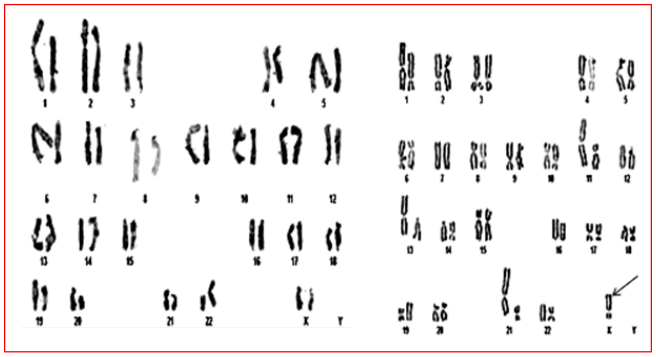

Figure 1: Normal karyotype with 46XX and Monosomy X with 45, X.

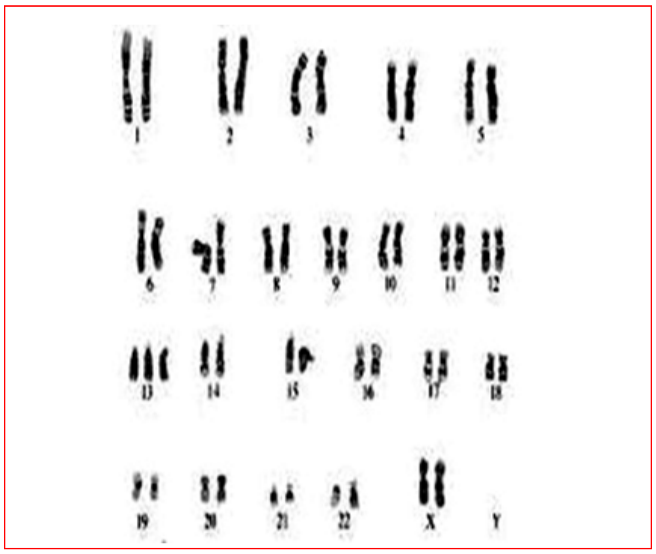

Figure 2: Chromosomes with trisomy 13 with 47XX respectively.

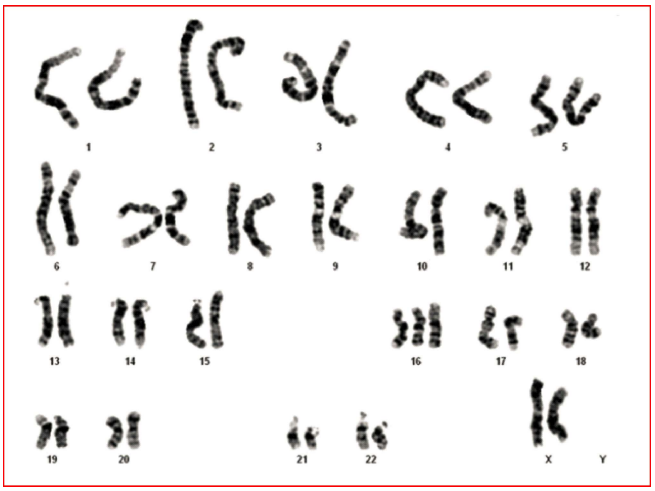

Figure 3: Chromosomes with trisomy 16 with 47XX respectively.

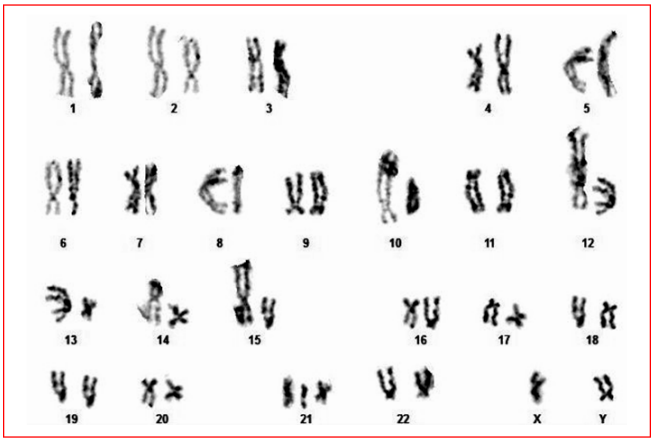

Figure 4: Chromosomes with trisomy 21 with 47XX respectively.

chromosomal anomalies in 35–50% of early pregnancy losses[11]. Trisomies were the most common abnormality (60.4%), with Trisomy 21 (42.3%), Trisomy 13 (34.6%), and Trisomy 16 (23.7%) being most frequently observed—findings that are in concordance with global cytogenetic data on early embryonic demise [12].

Our results underscore the recognized connection between increasing age of the mother and likelihood of meiotic nondisjunction. The highest frequency of trisomic losses was documented among women aged 30–34 years, consistent with the theory that age-related decline in oocyte quality predisposes to chromosomal segregation errors during meiosis I[13,14]. This is in line with prior research highlighting maternal age as the primary determinant of aneuploidy risk in assisted reproductive technologies as well as natural conception[14].

Contrastingly, Monosomy X, a common cause of embryonic lethality, was observed more frequently in younger women (mean age: 23.6 years). This suggests a divergent etiopathogenesis, possibly attributable to paternal sex chromosome loss, as previously reported in large-scale cytogenetic studies[15]. Polyploidy, accounting for 23.2% of the abnormal cases, was also found predominantly among women <30 years of age. Triploidy and tetraploidy, typically arising from dispermic fertilization or postzygotic errors, are universally fatal and remain a notable contributor to early gestational failure[15].

Structural chromosomal anomalies, though less frequent (9.3%), remain clinically significant due to their implications in recurrent pregnancy loss[16,17]. Balanced rearrangements such as reciprocal or Robertsonian translocations predispose to unbalanced gamete formation during meiosis, resulting in conceptuses with partial duplications or deletions incompatible with fetal

viability[16,17]. The presence of structural abnormalities in our cohort aligns with findings from similar Indian and international studies, which report parental chromosomal aberrations in 2–8% of couples with recurrent spontaneous abortions[18,19].

Moreover, a higher resolution analysis, such as chromosomal microarray, has demonstrated superior sensitivity in identifying submicroscopic copy number variants, particularly in cases with normal karyotype but phenotypic abnormalities[20]. This highlights the limitations of conventional G-banded karyotyping in fully elucidating genetic causes of miscarriage[20].

Limitations

This study is limited by its modest sample size and reliance on peripheral blood samples rather than direct analysis of products of conception, which may underrepresent mosaicism or placental chromosomal abnormalities. The exclusive use of conventional karyotyping limits detection to large-scale chromosomal rearrangements, potentially missing microdeletions, duplications, or epigenetic causes. Additionally, the absence of paternal chromosomal analysis precludes comprehensive evaluation of inherited structural abnormalities.

Conclusion

The findings of this study reinforce the central role of chromosomal anomalies—particularly trisomies, polyploidies, and structural abnormalities—in first-trimester spontaneous abortions. Maternal age remains a critical factor, especially for autosomal trisomies, whereas Monosomy X and polyploidy appear to follow distinct, age-independent mechanisms. Cytogenetic screening, especially in recurrent pregnancy loss, should be considered a pivotal component of clinical evaluation to guide future reproductive planning and genetic counseling. Advanced genomic techniques, including chromosomal microarrays or next-generation sequencing, may offer further insight into unexplained pregnancy losses.

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