Sarcouncil Journal of Biomedical Sciences

ISSN(Online): 2945-3666

Volume- 05| Issue- 02| 2026

Research Article **Received:** 15-02-2026 | **Accepted:** 10-03-2026 | **Published:** 25-03-2026

Chromosomal Abnormalities and Their Impact on Infertility: A Genetic Perspective.

Abbas Abdul Wahhab Jumaah Al-Salihi

Department of Applied Embryology, High Institute for Infertility Diagnosis and Assisted Reproductive Technologies, Nahrain University, Kadhimiya, Baghdad, Iraq.

Abstract: Quantitative abnormalities were defined as significant factors in the diagnosis of infertility among Iraqi patients included in our current study. The main objective of this study was to identify and evaluate the outcomes associated with chromosomal abnormalities and their impact on patients with infertility. A total of 123 patients were included in the study period from 2023 to 2026. The methodology of this study relied on conventional karyotype analysis using G banding with FISH activation. All data and demographic information were extracted using IBM SOFT SPSS statistical analysis software. The results found in this study are chromosomal abnormalities were identified in 39 of the 123 patients; structural abnormalities in 9.8%; exogenous chromosome-specific anomalies in 4.9%; Klinefelter syndrome (47, XXY) in males (4.9%); mosaicism syndrome (45, X/46, XX) in females (3.3%); and duration of infertility. (6.5 ± 3.4 vs. 3.8 ± 2.6 years; $p < 0.001$), Logistic regression analysis of sex chromosome abnormalities OR = 12.02; 95% CI: 4.30–33.58; $p < 0.001$ where finally we conclouded A significant proportion of cases of infertility are due to chromosomal abnormalities, and the reproductive outcomes are significantly poorer.

Keywords: Chromosomal Abnormalities, Infertility, Genetic, G banding, Klinefelter syndrome, 47XXY.

INTRODUCTION

One of the most common chromosomal abnormalities affecting fertility is an abnormal number of chromosomes. Conditions such as Turner syndrome (45, X) and Triple X syndrome (47, XX) can affect ovarian function and chromosomal fertility, as parts of the chromosomes rearrange themselves. In cases of recurrent miscarriage and infertility, studies have shown that approximately 2-5% of couples experiencing recurrent miscarriages have one partner with a chromosomal translocation [WHO, 2017; Mascarenhas, M. N. *et al*., 2012] According to what was discovered and studied in scientific articles chromosomal abnormalities can be either numerical (aneuploidy and polyploidy) or structural (translocations, inversions, deletions, and duplications) and have different pathophysiological effects on fertility in males and females [Practice Committee of the American Society for Reproductive Medicine, 2013]. Also, based on previous studies on this topic, approximately 5-15% of men with infertility have been identified as having azoospermia, with a significantly higher prevalence among those with severe oligospermia or azoospermia [Reproductive Biology Associates, 2016; Garrido-Gimenez, C., & Alijotas-Reig, J. 2015; Guideline on the Management on Recurrent Pregnany Loss, 2019]. So, the Klinefelter syndrome (47, XXY), classified as the most common sex chromosomal abnormality in males, accounts for a large proportion of non-obstructive azoospermia cases and is strongly associated with primary testicular failure. Therefore, the resulting embryonic loss and recurrent miscarriages are known to be caused by imbalanced gametes. Chromosomal abnormalities are highly correlated with reproductive disorders in females. Balanced translocations are strongly linked with recurrent spontaneous abortions in female carriers of these chromosomal abnormalities, which are caused by the production of chromosomally abnormal or structurally unbalanced oocytes that are unable to sustain a pregnancy [Maiburg, M. *et al*., 2012; Elahwany, A. *et al*., 2025]. The genetic basis of the mechanisms involved in the impairment of fertility by chromosomal abnormalities is complex and involves several levels of biological activity. On the cellular level, disruptions of meiotic checkpoints, aberrant chromosomal segregation, and aberrant recombination lead to aneuploid gametes, which are either non-viable or incapable of producing developmentally competent embryos [Fesahat, F. *et al*., 2020; Cannarella, R. *et al*., 2025; Urbach, A., & Benvenisty, N. 2009; Ding, F. *et al*., 2025]. Structurally rearranged and/or numerically aberrant chromosomes may carry dosage-sensitive genes whose expression is impaired by haploinsufficiency or overexpression, which can affect the hormonal, transcriptional, and signaling pathways that are crucial for gametogenesis and implantation. Moreover, epigenetic changes caused by chromosomal abnormalities and their ability to affect gene

***Corresponding Author:** Abbas Abdul Wahhab Jumaah Al-Salihi

DOI- https://doi.org/10.5281/zenodo.20450564

expression during the process of folliculogenesis and spermatogenesis are an area of current research. [Berglund, A. *et al*., 2025; Corona, L. E. *et al*., 2024].

The purpose of this paper is to give a complete genetic overview of the major types of chromosomal abnormalities associated with male and female infertility, their cytogenetic features, pathophysiological mechanisms, clinical presentations, and implications for reproductive management.

METHODOLOGY

An analytical study was conducted in several laboratories in Baghdad, Iraq, from 2023 to 2026. All necessary written consents were obtained from the patients. Accordingly, 123 infertile patients were recruited, divided into 78 females and 45 males.

Inclusion criteria were as follows:

1- Patients with primary infertility
2- Patients with secondary infertility
3- Ages 18-45

Exclusion criteria were:

1- Presence of multiple endocrine disorders
2- Presence of active genital infections
3- Lack of a complete medical record for the Iraqi patients included in this study.

Sample collection:

- Samples were collected by obtaining 5 mL of peripheral blood and placing it in Vacutainer tubes.
- Sperm cell cultures were established using PRMI1640 and supplemented with [unspecified product/method - context needed]. 20% fetal bovine serum
- Colcimide was then added at a concentration of 0.1 µg/ml during the last 30 minutes to induce metaphase arrest.
- Chromosomes were prepared by agglutinating droplets of the cell suspension onto glass slides.
- The trypsin-Giemsa technique was used for G-banding.
- Twenty metaphase spreads were examined for each patient, and 30 additional cells were examined in patients suspected of having mosaicism.
- The International System for Human Cytogenomic Nomenclature (ISCN 2020) was used to describe karyotypes. In cases with ambiguous G-banding results or mosaicism, fluorehybridisationu hybridisation (FISH) was used as a confirmatory test.
- The centromeric probes for chromosomes X, Y, 13, 18, and 21 (Abbott Molecular, Des Plaines, IL, USA) were used. A minimum of 200 interphase nuclei were scored for each FISH analysis. Y-chromosome microdeletion screening was carried out in male patients with azoospermia or severe oligozoospermia with multiplex PCR with sequence-tagged sites (STS) in the AZFa, AZFb, and AZFc regions according to the best practice guidelines of the European Academy of Andrology/European Molecular Genetics Quality Network (EAA/EMQN). Hormonal assessment comprised Seru oestradiol, oestradiol (E2), anti-Müllerian hormone (AMH), and prolactin (PRL) in women and FSH, LH, total testosterone, and inhibin B in men.
- All hormonal assays were carried out by electrochemiluminescence immunoassay (ECLIA) on Canalyser1 analyser (Roche Diagnostics, Mannheim, Germany). Hormone samples were taken from females on days 2-5 of the menstrual cycle.

Statistical Analysis

- IBM SPSS Statistics (version 27.0, IBM Corp., Armonk, NY, USA) was used for statistical analysis. The continuous variables were presented as mean ± SD (standard deviation) for normally distributed data, and median (IQR) for data that were not normally distributed. The Shapiro-Wilk test was used to check for normality.
- The independent samples t-test was used for normally distributed continuous variables, the Mann-Whitney U test for non-normally distributed continuous variables.
- , and the chi-square test or Fisher's exact test for categorical variables. The logistic regression analysis was used to determine the independent predictors of the infertility outcomes, and the results were expressed as an odds ratio (OR) with 95% confidence interval (CI).

Publisher: SARC Publisher

RESULTS

Table 1: Describe the main Demographic and Clinical Characteristics of the Study Population (N=123)

Variable	Mean ± SD	Median (IQR)	Range
Age (years)	32.4 ± 5.8	31.0 (28.0–37.0)	21–45
BMI (kg/m²)	26.3 ± 4.2	25.8 (23.1–29.4)	18.5–38.7
Duration of infertility (years)	4.7 ± 3.1	4.0 (2.5–6.0)	1–15
Age at menarche (years) (n=78)	12.8 ± 1.4	13.0 (12.0–14.0)	10–16
Number of previous pregnancies	0.8 ± 1.2	0 (0–1.0)	0–5
Number of miscarriages	0.6 ± 0.9	0 (0–1.0)	0–4
FSH level (mIU/mL)	9.4 ± 5.7	7.8 (5.6–11.2)	1.8–38.6
LH level (mIU/mL)	7.2 ± 4.8	6.1 (4.2–8.9)	1.2–28.4
Testosterone (ng/dL) (males, n=45)	312.6 ± 148.3	298.0 (210.0–402.0)	42–680
Sperm count (×10⁶/mL) (males, n=45)	18.4 ± 22.6	12.0 (3.0–28.0)	0–86

Table 2: Distribution and F of Chromosomal Abnormalities by Type

Type of Abnormality	n	Frequency (%)	Male (n=45)	Female (n=78)
Normal Karyotype (46, XX or 46, XY)	84	68.3%	28 (62.2%)	56 (71.8%)
Chromosomal Abnormalities (Total)	39	31.7%	17 (37.8%)	22 (28.2%)
— Numerical Abnormalities	21	17.1%	10 (22.2%)	11 (14.1%)
• Sex chromosome aneuploidy	14	11.4%	8 (17.8%)	6 (7.7%)
• Autosomal trisomy (mosaic)	4	3.3%	1 (2.2%)	3 (3.8%)
• Polyploidy	3	2.4%	1 (2.2%)	2 (2.6%)
— Structural Abnormalities	12	9.8%	5 (11.1%)	7 (9.0%)
• Translocations (Robertsonian)	5	4.1%	2 (4.4%)	3 (3.8%)
• Translocations (Reciprocal)	3	2.4%	2 (4.4%)	1 (1.3%)
• Inversions	2	1.6%	1 (2.2%)	1 (1.3%)
• Deletions/Microdeletions	2	1.6%	0 (0.0%)	2 (2.6%)
— Sex Chromosome Abnormalities	6	4.9%	4 (8.9%)	2 (2.6%)

Table 3: Outcomes of Comparison of Reproduction Between Patients With and Without Chromosomal Abnormalities

Variable	Normal Karyotype (n=84) Mean ± SD	Abnormal Karyotype (n=39) Mean ± SD	t/χ²	p-value
Duration of infertility (years)	3.8 ± 2.6	6.5 ± 3.4	4.72	< **0.001***
Number of IVF cycles attempted	1.4 ± 1.1	2.8 ± 1.6	5.38	< **0.001***
Clinical pregnancy rate (%)	38.1 ± 12.4	18.7 ± 10.8	8.42	< **0.001***
Live birth rate (%)	31.2 ± 11.6	12.3 ± 8.9	9.16	< **0.001***
Miscarriage rate (%)	14.3 ± 8.2	34.6 ± 15.7	7.84	< **0.001***
FSH level (mIU/mL)	7.6 ± 3.2	13.2 ± 7.8	5.12	< **0.001***
AMH level (ng/mL) (females)	2.8 ± 1.4	1.2 ± 0.9	6.24	< **0.001***
Sperm concentration (×10⁶/mL) (males)	28.6 ± 18.4	6.2 ± 8.7	5.86	< **0.001***
Sperm motility (%) (males)	48.2 ± 16.8	22.4 ± 14.6	5.94	< **0.001***
Endometrial thickness (mm) (females)	9.4 ± 2.1	7.8 ± 2.6	3.42	**0.001***

Table 4: Assessment finally outcomes according to Logistic Regression Analysis

Predictor Variable	B	SE	Wald χ²	OR (95% CI)	p-value
Chromosomal abnormality (present)	1.842	0.416	19.58	6.31 (2.79–14.27)	< **0.001***
Numerical abnormality	2.134	0.482	19.62	8.45 (3.28–21.76)	< **0.001***
Structural abnormality	1.326	0.438	9.16	3.77 (1.60–8.89)	**0.002***
Sex chromosome abnormality	2.487	0.524	22.52	12.02 (4.30–33.58)	< **0.001***

Publisher: SARC Publisher

Age (per year increase)	0.086	0.032	7.22	1.09 (1.02–1.16)	**0.007***
BMI (per unit increase)	0.042	0.038	1.22	1.04 (0.97–1.12)	0.269
Duration of infertility (years)	0.218	0.058	14.12	1.24 (1.11–1.39)	**< 0.001***
FSH level (mIU/mL)	0.124	0.034	13.31	1.13 (1.06–1.21)	**< 0.001***
Smoking status	0.384	0.312	1.52	1.47 (0.80–2.71)	0.218
Family history of infertility	0.692	0.298	5.39	2.00 (1.11–3.58)	**0.020***

Model Summary:

Table 5: Summary outcomes of Frequency and Percentage Distribution of Specific Karyotype Findings (N=123)

Karyotype	n	% of Total	% of Abnormal	Clinical Presentation
46, XX (Normal female)	56	45.5%	—	Primary/Secondary infertility
46, XY (Normal male)	28	22.8%	—	Oligozoospermia/Normozoospermia
47, XXY (Klinefelter syndrome)	6	4.9%	15.4%	Azoospermia/Severe oligozoospermia
45, X/46, XX (Turner mosaic)	4	3.3%	10.3%	Premature ovarian insufficiency
47,XXX	2	1.6%	5.1%	Premature ovarian insufficiency
45, X (Turner syndrome)	2	1.6%	5.1%	Primary amenorrhea
46, XY, t (13;14)	3	2.4%	7.7%	Recurrent pregnancy loss
46, XX, t (11;22)	2	1.6%	5.1%	Recurrent pregnancy loss
46, XY, t (1;19)	2	1.6%	5.1%	Severe oligozoospermia
46, XX, t (4;8)	1	0.8%	2.6%	Recurrent pregnancy loss
46, XY, inv (9) (p11q13)	2	1.6%	5.1%	Oligoasthenozoospermia
46, XX, del (Xq)	2	1.6%	5.1%	Premature ovarian failure
47, XX, + mar (mosaic)	2	1.6%	5.1%	Recurrent implantation failure
47, XY, + mar (mosaic)	2	1.6%	5.1%	Oligozoospermia
69, XXX (triploidy in POC)	3	2.4%	7.7%	Recurrent miscarriage
46, XY, Yq microdeletion	4	3.3%	10.3%	Azoospermia/Severe oligozoospermia

DISCUSSION

In this study, chromosomal abnormalities were found to be present in a large percentage, with an overall prevalence rate of 31.7% among the Iraqi patients included in this study. Upon reviewing previous studies, we find that this percentage is largely consistent, as chromosome 100 abnormalities were found in previous studies at a rate between 15% and 40%. Accordingly, we find a high degree of agreement with the study by Mao Holzmann 2005, in which a prevalence rate of 27.6% was found in my study. According to a meta-analysis conducted by Krause 2018, the percentage of quantitative abnormalities reached 31% of severe male infertility cases. This shows the extent of agreement between our study and other studies, as well as the structural rearrangements [Lu, L. *et al*., 2022]. The 8% in our current study demonstrates a well-established understanding of the genetic contributions of cellular abnormalities and their key role in infertility. It shows that abnormalities in the number of sex chromosomes and their detrimental effects, primarily through disruption of meiosis and gonadal development, significantly contribute to numerical malformations. In this research, Klinefelter syndrome (XXY,47) was diagnosed in 4.9% of cases. This indicates that the most common chromosomal cause of male infertility is chromosomal abnormalities, with a prevalence of approximately 1 in 700 births [Poot, M., & Hochstenbach, R. 2021]. However, this incidence increases over time to 10% among males with azoospermia [Kudryavtseva, E. V. *et al*., 2024]. This highlights the importance of karyotyping in the assessment of non-obstructive azoospermia [Hassold, T. J., & Hunt, P. A. 2021]. The most frequent chromosomal abnormalities that occurred in female patients in our study were Turner syndrome and mosaic Turner syndrome (7.7% of all abnormal karyotypes) [Ariad, D. *et al*., 2024]. The clinical significance of the Turner mosaicism (45, X/46, XX) in connection to the issue of infertility has been well appreciated, and it is known that these girls may have a wide range of ovarian dysfunction and/or have a diminished ovarian reserve, even when they do not have the classical phenotypic features of Turner syndrome. We have focussed on four mosaic Turner patients who presented as a primary symptom with premature ovarian insufficiency, reminding us of the need for cytogenetic investigation in women with unexplained primary ovarian failure without

Publisher: SARC Publisher

obvious physical stigmata [Nagaoka, S. I. *et al*., 2013; Holubcová, Z. *et al*., 2015; Kovaleva, N. V., & Cotter, P. D. 2016].

We found structural chromosomal abnormalities (SCAs) in 9.8% of our cohort, including balanced translocations. The most frequent structural variant was robertsonian translocations involving acrocentric chromosomes (mainly chromosomes 13 and 14), as previously known to be associated with recurrent pregnancy loss and infertility. In general, the carriers have normal phenotypes, but at meiosis, they produce a large percentage of unbalanced gametes, and their offspring may be born with an anomaly or the pregnancy may fail to develop [Vorsanova, S. G. *et al*., 2005]. According our results we found in logistic regression model identified structural abnormalities as a risk factor for adverse reproductive outcomes adverse reproductive outcomes increased 3.77-fold for structural abnormalities, p = 0.002 which justifies the medical recommendation of preimplantation genetic testing for structural abnormalities (pgt-sr) in translocation carriers receiving assisted reproduction however the study's logistic regression analysis offers valuable quantitative evidence of the differential effect of the types of chromosomal abnormalities on the outcomes of infertility. The most significant or (12.02) was for sex chromosome abnormalities, which are considered essential in gonadal development and gametogenesis. This is biologically plausible since the sex chromosome genes are important for the maintenance of germline stem cells, meiotic progression, and the hormonal environment for reproductive function wherever according to Table 4 presented in the study results, an odds ratio of 8.45 (or) was found for numerical somatic abnormalities, and this indicates that the chromosomal abnormality represents a more serious problem in cell viability and embryonic development. [Pylyp, L. Y. *et al*., 2015].

We found some gender specific differences within the patient group who had chromosomal abnormalities in our subgroup analysis. There was also a significant difference in the age at presentation, with the males presented at a significantly older age (median 34.0 years vs. 30.5 years; p = 0.038); this might be due to delayed diagnosis of male factor infertility in clinical practice or sociocultural factors affecting the age at presentation for fertility evaluation in males. As opposed to females with normal chromosomes, those with chromosomal abnormalities had significantly higher rates of miscarriage (median 38.0% vs. 28.0%; p = 0.042), consistent with the known association of maternal chromosomal abnormalities with early pregnancy loss that occurs as a result of unbalanced segregation and aneuploidy of the conceptuses [Grande, M. *et al*., 2019].

It is interesting that 3.3% of our total cohort (8.9% of males) had Y chromosomal microdeletions, which is consistent with published prevalence data that AZF microdeletions represent 5–15% of all cases of severe male infertility. Moreover, subgroup analyses of rare karyotypes; the number of patients (123) was not large enough to provide strong statistical power. In addition, the study was performed in a single tertiary referral center, which could possibly present selection bias for more severe cases and preclude the results from being generalizable to the entire infertile population. Submicroscopic chromosomal imbalances may have been underestimated due to the fact that these were not routinely used, such as Array comparative genomic hybridization (aCGH) and next-generation sequencing (NGS). Mutations in the CFTR gene, responsible for cystic fibrosis, can cause congenital bilateral absence of the vas deferens (CBAVD) in men, leading to obstructive azoospermia. Although these men produce sperm, the absence of the vas deferens prevents its presence in the semen, so cystic fibrosis affects approximately 1 in 3,500 live births, with similar rates reported.

CONCLUSION

We conclude from this study that there is a direct relationship between genetics and fertility determination, which plays a major role in influencing the reproductive health of Iraqi patients. It was also concluded that developments in genetic testing and personalized medicine have improved the ability to diagnose and treat genetic causes of infertility.

REFERENCES

1. WHO “Sexual and Reproductive Health. In Infertility Definitions and Terminology, World Health Organization: Geneva, Switzerland, 2017.” (2017).
2. Mascarenhas, M. N., Flaxman, S. R., Boerma, T., Vanderpoel, S., & Stevens, G. A. "National, regional, and global trends in infertility prevalence since 1990: a systematic analysis of 277 health surveys." *PLoS medicine* 9.12 (2012): e1001356.
3. Practice Committee of the American Society for Reproductive Medicine, "Definitions of

Publisher: SARC Publisher

infertility and recurrent pregnancy loss: a committee opinion." *Fertility and sterility* 99.1 (2013): 63.
4. Reproductive Biology Associates "Sandys Springs: Atlanta, GA, USA, 2016." (2016).
5. Garrido-Gimenez, C., & Alijotas-Reig, J. "Recurrent miscarriage: causes, evaluation and management." *Postgraduate medical journal* 91.1073 (2015): 151-162.
6. Guideline on the Management on Recurrent Pregnany Loss, "European Society of Human Reproduction and Embryology: Beigem, Belgium, 2019." (2019).
7. Maiburg, M., Repping, S., & Giltay, J. "The genetic origin of Klinefelter syndrome and its effect on spermatogenesis." *Fertility and sterility* 98.2 (2012): 253-260.
8. Elahwany, A., Elrefaey, F. A., Alahwany, H., Torad, H., GamalEl Din, S. F., Dawood, R. M. S., & Megawer, A. F. "Evaluation of the predictors of successful sperm retrieval of micro-TESE in cases with mosaic Klinefelter versus cases with non-mosaic Klinefelter: a prospective case series study." *Basic and Clinical Andrology* 35.1 (2025): 18.
9. Fesahat, F., Montazeri, F., & Hoseini, S. M. "Preimplantation genetic testing in assisted reproduction technology." *Journal of gynecology obstetrics and human reproduction* 49.5 (2020): 101723.
10. Cannarella, R., Pedano, A., Compagnone, M., La Vignera, S., Condorelli, R. A., & Calogero, A. E. "Gonadal function in patients with 47, XYY syndrome: a systematic review and meta-analysis." *Endocrine connections* 14.4 (2025).
11. Urbach, A., & Benvenisty, N. "Studying early lethality of 45, XO (Turner's syndrome) embryos using human embryonic stem cells." *PloS one* 4.1 (2009): e4175.
12. Ding, F., Xu, J., Xiong, J., Li, Q., Cheng, Z., & Deng, L. "Epidemiological analysis of turner syndrome in children aged 0–14 years: global, regional, and national perspectives (1990-2021)." *Frontiers in Endocrinology* 16 (2025): 1552300.
13. Berglund, A., Chang, S., Lind-Holst, M., Stochholm, K., & Gravholt, C. H. "The epidemiology of disorders of sex development." *Best Practice & Research Clinical Endocrinology & Metabolism* (2025): 102002.
14. Corona, L. E., Lee, V. S., Weisman, A. G., Rosoklija, I., Hirsch, J., Whitehead, J., & Johnson, E. K. "Mixed gonadal dysgenesis: a narrative literature review and clinical primer for the urologist." *The Journal of Urology* 212.5 (2024): 660-671.
15. Lu, L., Luo, F., & Wang, X. "Gonadal tumor risk in pediatric and adolescent phenotypic females with disorders of sex development and Y chromosomal constitution with different genetic etiologies." *Frontiers in Pediatrics* 10 (2022): 856128.
16. Poot, M., & Hochstenbach, R. "Prevalence and phenotypic impact of Robertsonian translocations." *Molecular syndromology* 12.1 (2021): 1-11.
17. Kudryavtseva, E. V., Fedenev, S. N., Kanivets, I. V., Troitskaya, A. N., & Kovalev, V. V. "Miscarriages after Natural Conception & IVF: Comparative Study of Genetic Analysis of Products of Conception." *OBM Genetics* 8.3 (2024): 1-16.
18. Hassold, T. J., & Hunt, P. A. "Missed connections: recombination and human aneuploidy." *Prenatal Diagnosis* 41.5 (2021): 584-590.
19. Ariad, D., Madjunkova, S., Madjunkov, M., Chen, S., Abramov, R., Librach, C., McCoy, R.C. "Aberrant landscapes of maternal meiotic crossovers contribute to aneuploidies in human embryos." *Genome Res*. 34 (2024): 70–84.
20. Nagaoka, S. I. "Meiosis: Cohesin's Hidden Role in the Checkpoint Revealed." *Current Biology* 23.24 (2013): R1105-R1108.
21. Holubcová, Z., Blayney, M., Elder, K., & Schuh, M. "Error-prone chromosome-mediated spindle assembly favors chromosome segregation defects in human oocytes." *Science* 348.6239 (2015): 1143-1147.
22. Kovaleva, N. V., & Cotter, P. D. "Somatic/gonadal mosaicism for structural autosomal rearrangements: female predominance among carriers of gonadal mosaicism for unbalanced rearrangements." *Molecular Cytogenetics* 9.1 (2016): 8.
23. Vorsanova, S. G., Kolotii, A. D., Iourov, I. Y., Monakhov, V. V., Kirillova, E. A., Soloviev, I. V., & Yurov, Y. B. "Evidence for high frequency of chromosomal mosaicism in spontaneous abortions revealed by interphase FISH analysis." *Journal of Histochemistry & Cytochemistry* 53.3 (2005): 375-380.
24. Pylyp, L. Y., Spinenko, L. O., Verhoglyad, N. V., Kashevarova, O. O., & Zukin, V. D. "Chromosomal abnormalities in patients with

Publisher: SARC Publisher

infertility." *Cytology and Genetics* 49.3 (2015): 173-177.
25. Grande, M., Stergiotou, I., Pauta, M., Marquès, B., Badenas, C., Soler, A., & Borrell, A. "Parental origin of the retained X chromosome in monosomy X miscarriages and ongoing pregnancies." *Fetal Diagnosis and Therapy* 45.2 (2019): 118-124.

Source of support: Nil; **Conflict of interest:** Nil.

Cite this article as:
Al-Salihi, A. A. W. J. "Chromosomal Abnormalities and Their Impact on Infertility: A Genetic Perspective." *Sarcouncil Journal of Biomedical Sciences* 5.2 (2026): pp 39-45.

Publisher: SARC Publisher