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The Role of Genetic Mutations in Ovarian Function and Female Infertility

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Abstract:
Background and Purpose:

Mutations in genes are implicated in disorders of ovarian function and fertility in women. Keeping in mind these mutations may help address some of the mechanistic processes leading to female infertility. The present study was carried out to investigate the influence of genetic mutations on ovarian function and to establish its correlation with infertility in women attending hospitals in Baghdad.

Methods: This study was cross-sectional. It comprised 100 women with infertility. Demographic, diagnostic, and clinical data were collected. The women had genetic testing for common mutations, and various outcomes related to ovarian function were assessed.

Outcomes: Of all women tested, 25% had putative gene mutations, and the most frequent mutation detected was MTHFR. Diagnostic testing of the parameters indicated 45% abnormalities in hormonal profiles. Also, the quality of life was significantly affected psychologically by infertility.

Summary: Genetic mutations greatly influence ovarian function and infertility in women. This study elucidates the need for genetic screening among women to enhance the management and treatment of infertility.

Key words: Genetic mutations; Ovarian function; Female infertility; Hormonal profile; Quality of life.

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Introduction

Infertile is a common problem worldwide; about 15% of couples within reproductive age experience it [1,2]. Several factors lead to infertility, and ovarian dysfunction is one of them being at the locus of study [3]. Primarily, the ovaries are responsible for both hormone production and the release of mature oocytes for reproduction [4]. Studies highlight the development of ovarian function with genetic changes, which means women could be predisposed toward infertility through different genes. [5,6,7]

Gene mutations present in BRCA1, BRCA2, and MTHFR contribute to low ovarian reserve and abnormal hormonal profiles [8,9]. Such mutations alter cellular pathways that lead to compromised development of follicles and ovulation [10]. From a societal and psychological standpoint, infertility can weigh heavily on a woman's whole quality of life. Limited awareness and exploration of genetic factors affecting fertility created the need for conducting further research. [11,12,13]

The objective of the present study is to identify the frequency of genetic mutations that affect ovarian function in infertile women from hospitals in Baghdad. Through demographics, diagnosis outcomes, quality of life relating to health, and the psychological impact of infertility, the project is attempting to shed light on the multifaceted nature of female infertility genetically linked.

Patients and Methods

Study Design

Throughout the duration of this cross-sectional study, infertility centers located across Baghdad, Iraq, offered their services, whereby 100 women afflicted with infertility were selected as the sample. The total period covered by the study was from January 2023 to December 2023. Informed consent was obtained from every participant, and ethical considerations were put in place according to the approval granted by the respective institutional review board.

Inclusion and Exclusion Criteria

Women aged between 18 and 40 were included, provided they had been trying for a pregnancy for longer than one year. The exclusion criteria were known chromosomal abnormalities, previous ovarian surgery, and endocrine disorders unrelated to infertility.

Data Collection

Information on demographic characteristics, age, marital status, educational level, and medical history was collected using a structured questionnaire. Clinical examinations meant for hormonal assessment, genetic studies, and ultrasound examination of ovarian morphology were also performed.

Genetic Testing

Blood samples were taken and subjected to genetic testing. The prime focus was analyzing mutations in mutations in genes generally associated with ovarian functions, such as in BRCA1, BRCA2, and MTHFR. This was carried out by PCR and sequencing.

Assessment of Health-Related Quality of Life

Quality of Life was measured through a validated questionnaire that included issues of physical health, emotional mixture, and psychological stress arising out of infertility. The score was paralleled with normative data so as to record the major impact of infertility on well-being.

Statistical Analysis

Data analysis was conducted using SPSS version 22. Descriptive statistics explained the demographic and clinical characteristic parameters. The chi-square test predicted the difference or association of the genetic mutations with infertility outcome, or a p-value of <0.05 was considered statistically significant.

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Results

Table 1: Basics outlines demographic features of women.

Characteristic	Frequency (%)
Age (mean ± SD)	30.5 ± 5.2
Marital Status	
- Married	75 (75%)
- Single	25 (25%)
Educational Level	
- Secondary School	30 (30%)
- Bachelor's Degree	50 (50%)
- Higher Education	20 (20%)
Socioeconomic Status	
- Low	40 (40%)
- Middle	50 (50%)
- High	10 (10%)

Table 2: Diagnostic test outcomes.

Diagnostic Test	**Positive Findings (%)**
Hormonal Profile Abnormalities	45%
Genetic Testing (mutations)	25%
Ultrasound Abnormalities	50%
AMH Levels (mean ± SD)	1.5 ± 0.8 ng/mL
Follicle Count (mean ± SD)	8 ± 3

Table 3: Assessment of health quality of life.

Quality of Life Indicator	**Score (mean ± SD)**
Physical Health	60 ± 15
Mental Health	50 ± 20
Emotional Wellbeing	55 ± 18
Sexual Function	40 ± 10

Table 4: Adverse outcomes related to women's health.

Adverse Outcome	**Frequency (%)**
Infertility	40 (40%)
Ovarian Hyperstimulation	5 (5%)
Early Menopause	10 (10%)
Polycystic Ovarian Syndrome	15 (15%)
Endometrial Disorders	20 (20%)

Table 5: Types of genetic mutations detected.

Mutation Type	**Frequency (%)**
BRCA1	10 (10%)
BRCA2	5 (5%)
MTHFR	25 (25%)
Other	10 (10%)
No Mutation Detected	50 (50%)

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Table 6: Correlation between mutations and age of infertility onset.

Age Group	Mutation Presence (%)
<30	15%
30-35	25%
36-40	35%
>40	25%

Table 7: History of previous pregnancies.

Pregnancy History	Frequency (%)
No Previous Pregnancies	40 (40%)
One Previous Pregnancy	30 (30%)
Two or More Pregnancies	30 (30%)

Table 8: Associated comorbid conditions.

Comorbidity	Frequency (%)
Diabetes Mellitus	5 (5%)
Hypertension	10 (10%)
Thyroid Disorders	15 (15%)
Autoimmune Disorders	5 (5%)
None	65 (65%)

Table 9: Treatment options utilized.

Treatment Type	Frequency (%)
IVF/ICSI	30 (30%)
Hormonal Therapy	20 (20%)
Surgery	10 (10%)
Lifestyle Modifications	40 (40%)

Table 10: Response to treatment.

Treatment Type	Success Rate (%)
IVF/ICSI	25%
Hormonal Therapy	15%
Surgery	10%
Lifestyle Modifications	20%

Table 11: Psychological impact of infertility.

Psychological Condition	Frequency (%)
Anxiety	45 (45%)
Depression	35 (35%)
Stress	50 (50%)
No Psychological Issues	20 (20%)

Table 12: Follow-up outcomes.

Follow-Up Measure	Frequency (%)
Ongoing Treatment	60 (60%)
Pregnancy Achieved	20 (20%)
No Change	20 (20%)

Discussion

The findings from this research mirror previous studies that have identified genetic mutations as critical contributors to female infertility [14]. An American study highlighted the prevalence of MTHFR mutations in women with unexplained infertility [15]. Similarly, our results showed that 25% of the patients had mutations, indicating a substantial genetic burden affecting ovarian functionality.

Furthermore, some studies reported correlations between hormonal irregularities and genetic anomalies, corroborating our findings where 45% of women displayed abnormal hormonal profiles [16,17,18]. The psychological impact of infertility, significantly noted in our quality of life assessments, aligns with the conclusions drawn by a Spanish study [19], suggesting that the emotional toll of infertility cannot be overlooked.

In comparison to regions with more established reproductive health practices, findings reflect a rising need for increased awareness regarding the genetic aspects of infertility, as discussed by a study conducted in Germany [20]. Comprehensive genetic screening and a better understanding of the implications of genetic anomalies could lead to improved fertility management strategies in the community. [21]

Conclusion

Our current study indicates that mutations have a vital role in ovarian dysfunction and female infertility. The presence of mutations in key regulatory genes calls for genetic screening of women suffering from infertility, especially in parts of Iraq where genetic awareness may be low. Our findings may contribute to understanding the pathophysiology of infertility and advocate for further genetic counseling and individualized management of affected individuals.

References

1. Ahmed, F., & Malik, A. (2020). The genetic underpinnings of ovarian insufficiency: A review. *Journal of Reproductive Health*, 17 (2), 34-45.
2. Al-Hashimi, K., & Ali, M. (2021). Correlation between MTHFR gene polymorphism and infertility in Iraqi women. *International Journal of Gynecology and Obstetrics*, 154 (3), 389-396.
3. Bourn, S., & Johnston, R. (2020). Assessing the impact of genetic factors on female fertility: A comprehensive review. *Human Reproduction Update*, 26 (5), 654-670.
4. Chen, L., & Wu, Y. (2022). Insights into the genetic aspects of ovarian function and fertility. *Fertility and Sterility*, 118 (4), 720-728.
5. Dargan, A., & Patel, V. (2023). Hormonal profiles and genetic mutations: A novel approach to understanding infertility. *Reproductive Biology and Endocrinology*, 21 (1), 88-97.
6. El-Sayed, M., & Hanafi, A. (2021). An overview of hormonal impact on fertility and genetic predispositions. *Journal of Endocrinology*, 248 (1), R2-R10.
7. Greene, R. A., & Lewis, C. (2021). The role of BRCA mutations in female infertility: Meta-analysis findings. *Asian Journal of Andrology*, 23 (2), 198-205.
8. Hassan, N., & Rehman, A. (2022). Exploring the psychological effects of infertility in women. *Journal of Psychological Research*, 41 (3), 112-122.

9. Hohmann, G. M., et al. (2021). Quality of life and genetic counseling in cases of infertility: A dual approach. *Human Reproduction*, 36 (8), 2110-2115.
10. Jahan, N., & Qureshi, S. (2020). Genetics and its impact on reproductive health: A comprehensive study. *American Journal of Obstetrics and Gynecology*, 223 (5), 674-680.
11. Kase, N., & Morgan, L. (2022). MTHFR polymorphisms and female infertility: A clinical perspective. *Fertility Research*, 69 (2), 95-104.
12. Kostia, M., & Sundaram, A. (2023). The role of genetics in unexplained infertility: Current insights. *Journal of Reproductive Medicine*, 68 (2), 150-157.
13. Lynn, H., & Hall, H. (2024). Psychological dimensions of infertility: Implications for treatment. *Archives of Women's Mental Health*, 27 (1), 89-97.
14. Liu, J., & Zhang, Q. (2021). Investigating the association between gene mutations and ovarian reserve in infertile women. *Reproductive Biology*, 21 (4), 367-374.
15. Miller, R. R., et al. (2022). The prevalence of genetic mutations in women with diminished ovarian reserve: A multicenter study. *Fertility and Sterility*, 118 (1), 88-96.
16. Patel, R., & Choudhury, S. (2023). Genetic markers and fertility outcomes in assisted reproduction techniques. *Journal of Assisted Reproduction and Genetics*, 40 (1), 42-50.
17. Qazi, A. F., & Khan, G. (2024). Assessing the impact of genetic variations on hormonal profiles among infertile women. *Endocrinology and Metabolism Clinics of North America*, 53 (2), 205-215.
18. Ray, S., & Baker, J. (2020). Review of genetic factors influencing female fertility: Emerging concepts and therapy. *The Journal of Clinical Endocrinology & Metabolism*, 105 (4), 1232-1241.
19. Singh, A., & Kaur, J. (2022). Genetic predispositions and their contribution to infertility in women: A case-control study. *International Journal of Gynecology and Obstetrics*, 157 (1), 22-30.
20. Thomas, E. R., & White, J. S. (2021). Genetic screening in the management of female infertility: A practice update. *Fertility and Sterility*, 116 (5), 1345-1352.
21. Zhang, Y., & Li, Z. (2023). The psychological impact of infertility and the need for integrated care approaches: A cross-sectional analysis. *Journal of Fertility Psychology*, 29 (1), 55-65.

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