INDIAN FERTILITY SOCIETY

# SIG Newsletter

November 2024

## Applied Genetics



## Role of Genetics in Infertility


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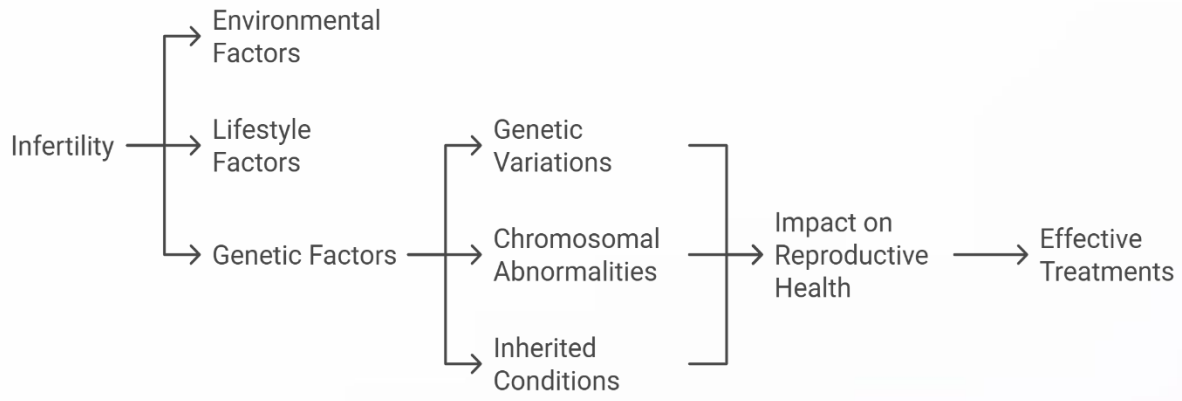
Infertility is a global public health concern, affecting an estimated 48.5 million couples worldwide, and its causes can be multifaceted, including environmental, lifestyle with a significant portion of cases being attributed to genetic factors. In both developed and developing countries, infertility has far-reaching social, psychological, and economic implications, particularly for women who often face stigma and discrimination.[1]

There are two forms of infertility, primary and secondary. Primary infertility arises from disruptions in germ cell development or function, leading to germ cell arrest and death. For women, primary infertility may manifest as conditions such as premature ovarian failure, polycystic ovary syndrome, endometriosis, and uterine fibroids. In men, primary infertility impairs spermatogenesis, often presenting with abnormal semen parameters. In contrast, secondary infertility is caused by factors that compromise the functional competence of mature gametes, such as impaired sperm motility or oocyte quality.[2]




## Epidemiology of Infertility

The prevalence of infertility has been steadily rising, with the age-standardized rate increasing by 0.37% per year for females and 0.29% per year for males globally. The highest prevalence is reported among individuals aged 35-39 years.[3] Infertility can be attributed to a combination of physical, lifestyle, genetic, psychological, and hormonal factors, with the male factor contributing to up to 50% of cases.[4]

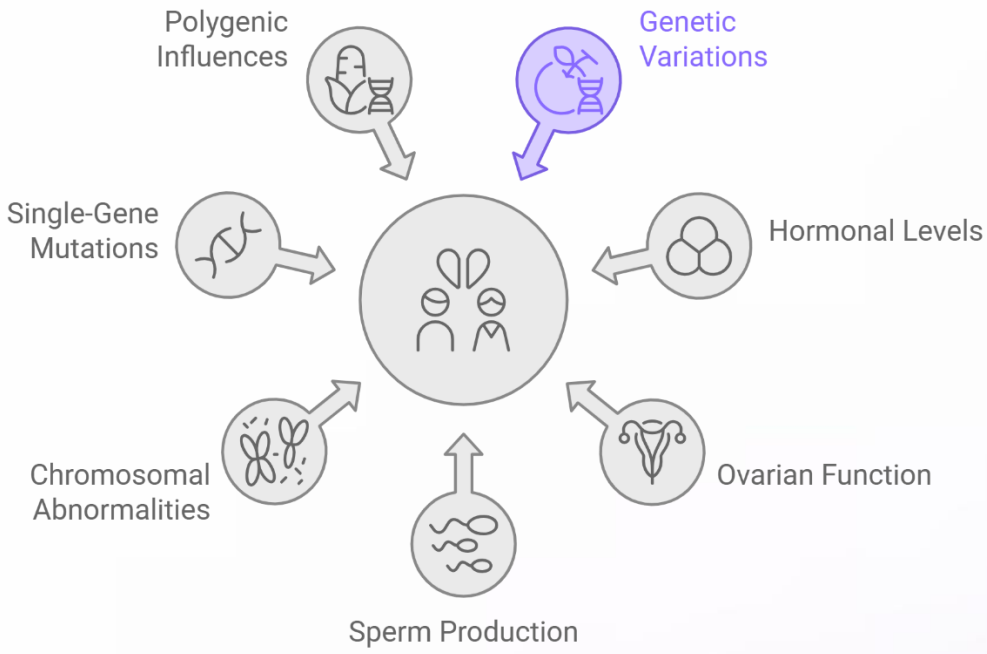
## Etiology

The etiology of infertility is often complex and multifactorial, involving a combination of genetic, biological, lifestyle, and environmental factors. Oxidative stress, resulting from an

imbalance between oxidants and reductants, is another common molecular mechanism underlying impaired sperm function and delayed pregnancy. Additionally, stress-induced hormonal changes, including alterations in cortisol, catecholamines, and reproductive hormones, have been associated with infertility.[5]

## Genetic Factors in Infertility

Genetic factors are less studied but play a crucial role in both male and female infertility. Chromosomal abnormalities, single-gene mutations, and complex polygenic influences can all contribute to reproductive difficulties. In men, genetic factors are associated with sperm abnormalities, such as impaired motility, morphology, and DNA integrity. Specific gene variants have been linked to azoospermia, oligozoospermia, and teratozoospermia. Similarly, in women, genetic factors can lead to ovarian dysfunction, polycystic ovarian syndrome, endometriosis, and other reproductive disorders.[6]




## Chromosomal Abnormalities

Chromosomal abnormalities, such as numerical (e.g., aneuploidy) and structural (e.g., inversions, translocations) variations, are a common cause of infertility.

Klinefelter syndrome (47,XXY): One of the primary concerns associated with Klinefelter syndrome is its impact on male fertility. It has been identified as the most persistent karyotype abnormality in infertile men, as it is related to the disruption of normal spermatogenesis, which can lead to azoospermia.

Y-Chromosome Microdeletion (YCMD) : Male infertility is a complex issue, and one of the underlying factors that can contribute to it is the presence of microdeletions on the Y-chromosome. YCMD can be found in 10-15% of men with azoospermia or severe oligospermia, which are the most severe forms of male infertility characterized by the absence or severely reduced sperm count. These microdeletions typically occur in three non-overlapping regions of the long arm of the Y-chromosome, known as AZFa, AZFb, and AZFc, which appear to contain multiple genes essential for spermatogenesis.[7]

## Monogenic Disorders

Single-gene mutations can also contribute to infertility. Examples include mutations in the CFTR gene, which can cause congenital bilateral absence of the vas deferens, and mutations in the INSL3 and RXFP2 genes, which have been associate with cryptorchidism and impaired spermatogenesis.

## Polygenic Disorders

Infertility can also result from complex, polygenic interactions involving multiple genes and environmental factors. Conditions such as endometriosis, polycystic ovarian syndrome, and male factor infertility often have a strong genetic component, with numerous genetic variants contributing to the overall risk.

## Epigenetics

Recent research has also highlighted the role of epigenetic modifications in infertility. Epigenetic changes, such as DNA methylation, histone modifications, and microRNA regulation, can influence gametogenesis, embryonic development, and reproductive outcomes without altering the underlying DNA sequence.

## Environmental Factors

In addition to genetic factors, environmental exposures, such as chemicals, radiation, and lifestyle factors, can also impact fertility. Heavy metal exposure, air pollution, and endocrine-disrupting chemicals have all been linked to reduced sperm quality and female reproductive disorders.

## References


1. Sharma, S R., Mittal, S., & Aggarwal, P. (2009, September 4). Management of infertility in low resource countries. Wiley, 116(s1), 77-83. https://doi.org/10.1111/j.1471-0528.2009.02311.x
2. Vialard, F., Mandon-Pepin, B., Pellestor, F., Ziyyat, A., Albert, M., Molina-Gomes, D., Selva, J., & Fellous, M. (2009, March 1). Anomalies génétiques et infertilité masculine. Springer Nature, 19(1), 2-16. https://doi.org/10.1007/s12610-008-0002-y
3. Karunyam, B V., Karim, A., Mohamed, I N., Ugusman, A., Mohamed, W., Ahmad, M F., Abu, M A., & Kumar, J. (2023, June 29). Infertility and cortisol: a systematic review. Frontiers Media, 14. https://doi.org/10.3389/fendo.2023.1147306
4. Pinto, F M., Larrañaga, A O., Candenas, L., & Subirán, N. (2023, April 10). The Role of Sperm Membrane Potential and Ion Channels in Regulating Sperm Function. Multidisciplinary Digital Publishing Institute, 24(8), 6995-6995. https://doi.org/10.3390/ijms24086995
5. Agarwal, A., & Majzoub, A. (2016, November 28). Free Radicals in Andrology. Springer International Publishing, 1-21. https://doi.org/10.1007/978-3-319-41749-3_1
6. O'Reilly, E K., Sevigny, M., Sabarre, K., & Phillips, K P. (2014, October 14). Perspectives of complementary and alternative medicine (CAM) practitioners in the support and treatment of infertility. BioMed Central, 14(1). https://doi.org/10.1186/1472-6882-14-394
7. Report on evaluation of the azoospermic male. (2006, October 19). Elsevier BV, 86(5), S210-S215. https://doi.org/10.1016/j.fertnstert.2006.08.030



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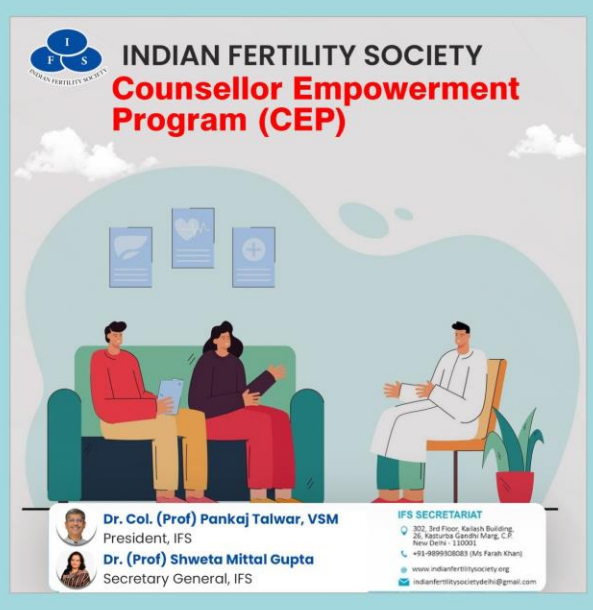
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