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**Review Article**

# What is The Prevalence of Genetic Disorders That Cause Effects On In Fertility?

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## ABSTRACT

Infertility rates increases day by day and major reason can be genetic disorders. **Objective:** To review the prevalence and effects of genetic defects that can cause infertility. 10 to 20% of patients have severe infertility disorders. To find out whether genetic material defects are really major infertility cause or not? And what are their prevalence. Infertility is common health condition. Genetic cause are majorly unexplained but 2300 genes expressed that can retranslate and their mutation can cause of infertility. Genetics and infertility have major relationship due to maternal age effect. Handful of genes and genetic effects are associated with human genome to express genes. Infertility controls by Genetics can be treated with low risk that is very effective. Mostly tests added to check male infertility annually. **Methods: Ar**ticles were searched from (1997-2011) by using Google scholar, PubMed etc. All data was reviewed from infertility complaints. The data was extracted from Medline, PubMed and Obsgyne online library. **Results:** Those studies which were conducted between years 1985 to 2011 were included in this review. The sample size included more than 300 men and women aged between 20 to 40 years. In this study we evaluated that genomic technologies and advent of fertility tips can help in fertile patients. Metabolic chromosomal defects can be genetic disorders by deletion of chromosomes abnormally. SOX9 chromosome is also play basic role in men fertility. **Conclusions:** Infertility can be caused by genomic abnormalities and chromosomal abnormalities. Female ovarian patterns have major genetic issues that cause infertility. Molecular techniques can help to reduce the abnormalities.

## INTRODUCTION

The complex disorder of male and female reproductive system is infertility. Male and female sterility depends upon two forms Primary and Secondary. If we see the physiology or cell structure of germ cells then we can understand that the primary form affects the germ cells which can cause ultimate death of of germ cell. In primary female infertility we can see that premature ovarian failure and endometriosis are basic sterility defects due to which female infertility can happen. On the other hand, primary male sterility basically directly affect spermatogenesis and also directly associated with abnormal sperm count sperm motility. Then the secondary infertility can occur due to syndrome genetic defects like endocrine development defects and metabolic defects. Those genetic syndromes that can cause female or male infertility are fragile X syndrome, Kartagener's syndrome etc. Other notable conditions include different disorders like reproductive digenesis disorder, hypogonadism and congenital bilateral absence of vas deferens [1,2]. Basic disruption of steroid synthesis and metabolism that are basically defects of CYP17, CYP21 AND CYP21A2 mutations [3-5]. These are genes mutations that can cause by endocrine defects mitochondrial mutations cause toxic defects that can also result of secondary male and female infertility [6]. Many genetic defects fall in following categories: chromosomes aberration, DNA copy no micro deletion and duplication single gene disorders and epigenetic disorders. When the infertile couples labelled with the descriptive diagnosis of infertility and they do not provided with cause of their disease, Although their disease can be diagnosed with advanced technology under the supervision by safety or ethical concerns so the advanced study in the failed of

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fertility realized in the 21st century with proper care and understanding of full procedure for this the scientist collected 400 mutant mouse models with reproductive phenotype that have proper reproductive gonadal stem cells than the virtual study has explosive results in which gene mutation and polymorphisms like causes build relationship with human infertility[2]. The regulation and secretion of sex hormones are resulted by hypothalamic-pituitary-gonadal axis. The neurons of GnRH relatively conduct inputs to the axis which generate integration in central nervous system before migrating to the hypothalamus the genetic defect that cause disjunction of hypogonadotropic hypogonadism is identified in some patients, when the experiences collected 70% mutation remain unidentified. Disruption of signals in the complex process of GnRH can cause neuronal signal disruption [7]. Infertility may cause due to the mutation in CFTR gene which can cause obstruction and other deficiency in maturation of sperm [8]. The extreme gonadal-mutations in sex can cause the reversal of chromosomes 46. It is observed that maternal tobacco consumption can cause severe effects on spermatogenesis [9-11]. The new study also indicated that endometrial-deposits can cause infertility but these are not the alone indication of infertility. The hundreds of genes pathways involved and the large equal options and opportunities are required which are leading to infertility. It is estimated that worldwide one in six couples face the incidence of diseases which is associated with infertility. It can affect both male and female depending upon the distribution of factors which is molecular or genetic effect underlying phenotypes of infertility [12].

## METHODS

In this systematic review data which is collected by PubMed, sciencedirect, biimedcentral.com and obs-gynae online library that were queried for studies published between 1985-2011 and also some research done until today on prevalence of Genetic disorders cause and effect on infertility. Data extracted from more than 10 articles. In thus systematic article we reviewed cross sectional style analytical studies for the collection of data.

## RESULTS

Those studies which were conducted between years 1985 to 2011 were included in review. The sample size included more than 300 men and women aged between 20 to 40 years. SOX9 chromosome is also play basic role in men fertility. Semen collection and measurements done and proper advances can be seen through research to follow up infertile patients. Comprehensive diagnosis started to advance the research and to increase the prognostic information.

## DISCUSSION

In 1985 research was conducted on Parental chromosomes abnormalities. this research was done by surveys of 79 couples with two or more pregnancy loses and have chromosomes abnormalities of 3.2%. In this survey the dominance of female to male was noted 50% of abnormalities caused by translocations sex chromosomal defects are majorly occurring in females which are about 12%. If the abnormal developments of neurons that produce GnRH occur the impair-action of genes developed which can cause deficiency of hormones. In translocation abnormality it is observe that abnormal zygote is found parents are normal in these abnormalities but pregnancies of these parents have high incidence of chromosomes abnormalities and spontaneous abortions. In these abnormalities parental karyotypes also examined and parental diagnosis also considered [13]. In 1994 research was conducted on Autosomal sex reversal and dysplasia cause by mutation and related gene. In this research it is found that chromosomal and expression found on SOX9 gene. This gene structurally effect on fetal testis and skeletal issue Non-translocation cases identified inactive mutation on one SOX allele this gene deficiency cause dysplasia and other gene defects. In 1997 the research was conducted on Mutation in SRY and SOX9. In this research it is observed that gene in Y chromosomes that causes defect of potential gonad defect that develop as a testis. This Y chromosome on the gene is named as testis determining factor. During the male meiosis, DTF can result XX male phenotype. The specific region of Y chromosome is reserved for SRY. This can cause mutation in XY female the tendency of SRY to develop sex reversal males is high. There are large numbers of SRY copies of protein that do not bind DNA. The DNA bending to bring different positions which facilitate transcription [14]. The disruption of endocrine which is occurring during pregnancy have important effects on fertility. The basic percentage of women age during the first baby birth and second baby birth is considered to have high parity between 35years [15]. The aging of ovaries can cause decline number of oocytes the sudden decrease of oocytes is observed that the age if 37. This can cause by Genetics or difficulty due to meiotic genetic abnormality [16]. The spermatogonia cannot survive to become the level of mature sperm maturation. It can cause high rate of genetic abnormalities that can occur due to BAX proteins deficiency. It is also considered that pituitary express genes that are FSHB and their receptors abnormality cause decrease number of spermatozoa. Many women have mutation that can cause POF due to FOXL2 gene [17-20]. In 2001, a research was conducted on decreased fertilization rate and embryo quality after oligozoospermic men with

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micro deletion in the ascosporic factor region. A total of 195 cases were monitored. Couples medical reports and hormones level were measured. Comparison between age and serum hormone was conducted. About 75% were oligospermic, 116 were azoospermic. The oligospermic patients had very low chances of pregnancy [21]. In 2002, a research was conducted on pregnancy and inherited metabolic disorders. It was observed that metabolic disorders can cause complications during pregnancies and can affect mothers and fathers as we manage metabolic disorders in childhood than the chances of pregnancy increase [22].

## CONCLUSION

Infertility can be caused by genomic abnormalities and chromosomal abnormalities. Female ovarian patterns are having major genetic issues that cause infertility. Molecular techniques can help to reduce the abnormalities. In reality we are unable to establish genotype phenotype relationship and various testicular patterns to see infertile men

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