Opinion
Volume 16:3, 2022

Journal of Molecular and Genetic Medicine

ISSN: 1747-0862 Open Access

Gene Polymorphism and Infertility in Women

Thomas Margret*

Department of Molecular Genetics, MolGen S Research Centre, UK

Introduction

Infertility in women is a serious medical issue with many causes unknown. It happens when implantation fails, a fertilized embryo dies after implantation, or the egg is unable to travel from the ovary to the uterus. The discovery of polymorphisms linked to a disease's pathogenesis can aid in the understanding of the disease's pathophysiology, and this knowledge can be used to improve the prognosis for women with a specific ailment, such as Polycystic Ovary Syndrome (PCOS). Because an altered response to ovarian stimulation is a symptom of the condition, understanding it's an etiology could aid in establishing the parameters that govern an individual's response to ovarian stimulation. Several genes involved in ovarian function and metabolism have been linked to an elevated risk of PCOS, but none of them is powerful enough to predict PCOS susceptibility or treatment response on its own. FSHR p.N680S, a single-nucleotide polymorphism in exon 10 of the FSH receptor (FSHR) gene, has repeatedly been found to have a substantial connection with ovarian responsiveness to FSH.

Description

Genetic association studies are performed to look for genes or genetic markers that are linked to a specific disease phenotype or trait. Natural variations or polymorphisms in the DNA sequence of people are identified and characterized in this research. If a connection exists, a particular variant will be seen more frequently in a person carrying the characteristic than would be predicted by chance. Depending on where they occur in relation to a gene, variants are divided into different groups. Variants might exist within or outside of a gene, for starters. Those found within a gene can be found in exons, introns, or regulatory regions. A gene's variants can be active or inactive. A phenotypic abnormality might be caused directly by functional variation within a gene, or it can enhance disease risk. Functional variants in coding areas can alter protein sequence, while functional variants in non-coding regions can affect RNA transcription and processing [1,2].

The two most common ways for identifying genes that cause disease when they are mutated are genetic and functional approaches. Genetic mapping methods are used in genetic approaches to the study of hereditary disease to pinpoint the site within the human genome of a genetic component impacting disorder development. The gene or genes in that region are next examined for signs of involvement in the illness. In functional approaches to genetic disease research, on the other hand, 'candidate genes' that may be mutated in individuals with the condition under investigation are discovered based on other data. Genetic techniques to disease gene identification were once thought to be less direct and time-consuming than functional approaches, which had some merit. Genetic techniques may now be used with continuously increasing efficiency to the study of an ever-expanding collection of disorders, because to recent breakthroughs in laboratory technology, analytic approaches, and available data (the latter including the almost entire sequence of the human genome). The use of genetic approaches has substantially aided our understanding of hereditary illnesses in recent years, and this fruitful use of genetic approaches is likely to continue.

The subsequent identification of the gene mutated in HD (i.e., the HD 'disease gene') by a collaborative group of 58 authors from six institutions took a decade and the work of many dozens of researchers affiliated with a variety of collaborating and competing laboratories. Identification of the disease's likely mode of inheritance; application of experimental and analytical methods to map the disease gene and then refine its location; identification of candidate genes; screening of candidate genes for mutation; and determination of the functional consequences of the mutation are typical steps in genetic mapping studies. The subsequent identification of the gene mutated in HD (i.e., the HD 'disease gene') by a collaborative group of 58 authors from six institutions took a decade and the efforts of many dozens of researchers affiliated with a variety of collaborating and competing laboratories. Identification of the disease's likely mode of inheritance; use of experimental and analytical methods to map the disease gene and then refine its location; identification of candidate genes; mutation screening of candidate genes; and determination of the functional consequences of the mutation are all common steps in genetic mapping studies. A disease gene can be mapped using a variety of experimental and analytical methods [3-5].

Conclusion

These methods rely on genotyping of genetic markers in the lab, albeit the number and type of markers used varies. Polymorphic simple sequence repeats have been the most utilized genetic markers in mapping investigations to date (SSRs). These are brief, tandemly repeated sequences that are widely dispersed. Dinucleotide and tetranucleotide repeats are the most often used SSRs. Because the number of copies of the repeat unit differs between alleles, allele length varies. The inheritance pattern can be determined by amplifying the repeat from genomic DNA with distinct primers flanking the repeat and electrophoresing the PCR products to segregate the alleles by size.

References

1. Coulam, Carolyn B., and R.S. Jeyendran. "Thrombophilic gene polymorphisms are risk factors for unexplained infertility." *Fertil Steril* 91 (2009): 1516-1517.
2. Ayvaz, Özge Üner and Evrim Ünsal. "Evaluation of in vitro fertilization parameters and estrogen receptor alpha gene polymorphisms for women with unexplained infertility." *J Assist Reprod Genet* 26 (2009): 503-510.
3. Wang, Wenwen and Shixuan Wang. "Association of an oestrogen receptor gene polymorphism in Chinese Han women with endometriosis and endometriosis-related infertility." *Reprod Biomed* 26 (2013): 93-98.
4. Chang, Cherry Yin-Yi, Hui-Wen Chang and Fuu-Jen Tsai. "MUC4 gene polymorphisms associate with endometriosis development and endometriosis-related infertility." *BMC Med* 9 (2011): 1-10.
5. Gimenes, Cristiane, Bianca Bianco and Caio Parente Barbosa. "The progins progesterone receptor gene polymorphism is not related to endometriosis-associated infertility or to idiopathic infertility." *Clinics* 65 (2010): 1073-1076.

***Address for Correspondence:** Thomas Margret, Department of Molecular Genetics, MolGen S Research Centre, UK; E-mail: margretmolg20@yahoo.com*

Received: 05-March-2022, Manuscript No: jmgm-22-61722; **Editor assigned:** 07-March-2022, PreQC No. P-61722; **Reviewed:** 12-March-2022, QC No. Q-61722; **Revised:** 17-March-2022, Manuscript No. R-61722; **Published:** 22-March-2022, DOI: 10.37421/1747-0862.22.16.542

How to cite this article: Thomas, Margret. "Gene Polymorphism and Infertility in Women." *J Mol Genet Med* 16 (2022): 542