IRJSE International Research Journal of Science & Engineering
Int. Res. J. of Science & Engineering, Special Issue A8, July, 2020
ISSN: 2322-0015
SJIF Impact Factor 6.70
RESEARCH ARTICLE
OPEN ACCESS

Study of Genotype Frequency Between PCOS and Non PCOS Womens in Vidarbha region

Lilhare MU and Pawar SS

Govt. Vidarbha Institute of Science and Humanities, Amravati-444604 (MS) India
*Corresponding author: E. Mail-

Manuscript Details

Available online on https://www.irjse.in
ISSN: 2322-0015

Editor: Dr. Arvind Chavhan

Cite this article as:
Lilhare MU and Pawar SS. Study of Genotype Frequency Between PCOS and Non PCOS Womens in Vidarbha region, *Int. Res. Journal of Science & Engineering*, 2020, Special Issue A8:65-68.

Article published in Special issue of International e-Conference on "Sustainable Development : A Biological and Socioeconomical Perspective" organized by Government Vidarbha Institute of Science and Humanities Amravati, Maharashtra, India date, June 26-27, 2020.

Abstract

A total 170 Vidarbhian women's were included in present study. Among them 65 were Non-PCOS (Non-Poly cystic ovarian syndrome) considered as control group and 105 were PCOS (Poly cystic ovarian syndrome) considered as experimental group. The PCOS women's were diagnosed based on the 2003 Rotterdam Criteria. Out of 105 PCOS women's 40 were found to carry Mutant Homozygous (AA), 50 were found to carry Mutant Heterozygous (AG) and 15 women's are Wild type Homozygous (GG). Out of 65 Non-PCOS women's 18 are found to carrying Mutant Homozygous (AA), 15 are found to carrying Mutant Heterozygous (AG) and 15 women's are found to carrying Wild type Homozygous (GG). By observing the present study the mutant form is highly elevated in PCOS women's that shows PCOS women's are slightly suffering with PCOS as compared to non-PCOS women's.

Keywords: *PCOS and Non PCOS Women, Genotype frequency, Vidrabha region.*

Introduction

Polycystic ovary syndrome (PCOS) is a genetically complex disorder characterized by amenorrhea or oligomenhorreal hyperandrogenemia. It is the most common endocrinopathy among women of reproductive age [1, 2].

Polycystic ovarian syndrome is a complex endocrine disorder occurring at a frequency of 11.2% in women of reproductive age group, out of which prevalence of PCOS in teenage girls may be as high as 50% [3].

It is the commonest cause of menstrual irregularities and infertility [4]. Young adolescent girls experience full range of symptoms from irregular menses, amenorrhea, menorrhagia, hirsutism, acne, skin pigmentation, alopecia, ovarian cysts. Other symptoms like anxiety, depression, thyroid problems and galactorrhea, may exist. Obesity or propensity to weight gain is a common feature. Characterized by chronic an ovulation, hyperandrogenism, hormonal imbalance and metabolic disorders [5].

In humans, the gene CYP19, located on chromosome 15q21.1, encodes the aromatase enzyme [6]. Aromatase catalyzes the final step of estrogen biosynthesis, the conversion of C19 steroids to estrogens. A single gene (CYP19) encodes aromatase, and is expressed in specific tissues, including the brain, gonads, fat, skin and the placenta [7]. In PCOS, activity of enzyme CytochromeP450 c17 (CYP17), which converts progesterone to 17-hydroxyprogesterone and from 17-hydroxyprogesterone to androstenedione (A4) is exaggerated and a decreased activity ofCYP19A1 favours androgen production on these women [8].

Materials and method

A total of 170 Vidarbhian women's were included in present study. Among them 65 were Non-PCOS considered as control group and 105 were PCOS considered as experimental group. The study included only patients diagnosed in Hospitals with PCOS phenotype, Patients were assessed by history and physical examination. History, after biodata is documented (name, age, address, occupation). First information regarding PCOS womens and related symptoms were collected from various pathological centers, beauty parlors and gynecological hospitals during the period of January 2014 to March 2018. All the women's were of Indian origin. The study was conducted at Vidarbha region, Maharashtra India.

The PCOS women's were selected on the basis of two diagnosis methods, clinical and biochemical **[9]**. We were selected women's those who are already diagnosed as a PCOS women's by Doctors as experimental group and non PCOS women's randomly selected on the basis of personal consent and information about normal health and age as control group.

Statistical Analysis

Genotype frequency was calculated by using Hardy-Weinberg equation. Hardy-Weinberg distribution of genotypes in the PCOS and non PCOS groups was assessed. Fisher's Exact Test was used to compare the CYP19 gene genotype distributions in the PCOS and non PCOS groups. The odd ratio (OR), its standard error and 95% confidence Interval is calculated according to Altman, 1991.

Result and Observation

The results of the genotype frequency, the differences in genotype distribution, and odds ratio, confidence interval, p value for each polymorphism are summarized in tables (Table 1). Distribution of different allelic forms and their corresponding values of significance are enumerated. It is evident that there is near to significant difference between the distribution of the genotypes between PCOS and non PCOS women.

Table 1 - The genotype frequencies of PCOS and non PCOS women's

Genotype	PCOS women's (105)	Non PCOS women's (65)	P value	Odd ratio	95% CI	Z – Statistic
AA	40 (0.383)	18(0.274)	0.0842	2.2222	0.8977 to 5.5012	1.727
AG	50 (0.471)	32(0.498)				
GG	15 (0.144)	15(0.226)				

The genotypic frequency of PCOS and non-PCOS women's was calculated by Hardy-Weinberg equilibrium. In 170 women's the CYP19 rs2414096 genotypic distributions were for AA, for AG and for GG.

Genotypic distributions (AA, AG and GG) in women with PCOS women's (0.38, 0.47 and 0.14, respectively) were near to significantly different from the non-PCOS women's (0.27, 0.49 and 0.22, respectively).

It was also observed that genotype frequency AA and GG shows highest in PCOS women's as compare to non PCOS women's. Nearly significant difference was observed in PCOS and non PCOS women's (p=0.08).

Discussion

It is a multifactorial endocrine disorder, which demonstrates menstrual disturbance, infertility and hyperandrogenism[10]. In view of strong evidence implicating the importance of CYP19 gene polymorphisms in androgen metabolic pathway. The genotypic frequency of PCOS and Non-PCOS women's were calculated by Hardy-weinberg equilibrium. In 170 women's the CYP19 rs2414096 genotypic distributions were for AA, for AG and for GG. Genotypic distributions (AA, AG and GG) in women with PCOS (0.38, 0.47 and 0.14 , respectively) were significantly different from the non-PCOS (0.27,0.49 and 0.22, respectively). Statistically significant difference was not observed between PCOS and Non PCOS (p=0.08).

By observing the present study the mutant form is highly elevated in PCOS women's, that shows PCOS women's are slightly suffering with PCOS as compared to Non-PCOS women's. Present study correlate with the[11].

The AA genotype in PCOS was significantly higher than that of the other two genotypes and this suggests that aromatase activity was augmented in the AA genotype [11]. Reduced aromatase activity may lead to ovarian hyperandrogenism and the development of PCOS, which can be deduced from the facts that a higher frequency of PCOS is observed in people with aromatase deficiency caused by rare loss-of-function mutations [12,13 ,14] and antral follicles taken from PCOS women exhibits no aromatase activity[15].

This shows higher frequency of alleles A and G are observed in PCOS women's as compared to non-PCOS women's. Petry *et al*.,[16] who found that the 'A' allele, which was more prevalent in precocious pubarche (PP) girls is associated with increased testosterone concentrations in both the Barcelona PP case-control study and the Oxford population study.

Conclusion

As compared to PCOS and non PCOS women's, we found that the PCOS shows higher frequency of mutant homozygous and heterozygous conditions as compared to non-PCOS women's.

Present study demonstrated that homozygous AA genotype confers significant protection towards PCOS over the other two genotype.

It is evident from genetic aspects were studied and it was found that genetic frequency of AA, AG and GG gene shows nearly significant result as compare to control group. This could be influenced by some environmental forces as well as inter-ethnic admixture, which could change allelic frequency.

Over all we found that alteration of genotype frequency in PCOS women have suggested that the variation in this gene is linked to the androgen excess in PCOS womens. And if androgen excess in womens shows number of complication in maintaining hormonal balances and their consequences as PCOS like syndrome appeared in womens of Vidarbha region.

Conflicts of interest: The authors stated that no conflicts of interest.

References

1. Diamanti-Kandarakis E., Kouli C.R., Bergiele A.T., Filandra F.A., Tsianateli C., Spina G.G.,*et al* (1999) A survey of the polycystic ovary syndrome in the Greek island of Lesbos: hormonal and metabolic profile. *Journal of Clinical Endocrinology and Metabolism*, 84:4006–4011.
2. Knochenhaver E.S., Key T.J., Kahsar-Millar M., Waggoner W., Boots L.R and R. Aziz (1998) Prevalence of polycystic ovary syndrome in unselected black and white woman of the south eastern United States: a prospective study. *J Clinc Endocrinol Metab,* 83: 3078-82
3. Coviello A.D., Legro R.S and A. Dunaif (2006) Adolescent girls with polycystic ovary syndrome have an increased risk of the metabolic syndrome associated with increasing androgen levels independent of obesity and insulin resistance. *J Clin Endocrinol Metabol*, 91(2):492-7.
4. Yildiz B.O, Knochenhauer E.S and R. Azziz (2008) Impact of obesity on the risk for polycystic ovary syndrome. *J Clin Endocrinol Metab.* 93(1):162-8.
5. Anjali Choudhary, Shweta Jain and Priyanka Chaudhari (2017)Prevalence and symptomatology of polycystic ovarian syndrome in Indian women: is there a rising incidence?*Int J Reprod Contracept Obstet Gynecol* ;6(11):4971-4975
6. Toda K and Y. Shizuta (1993) "Molecular cloning of a cDNA showing alternative splicing of the 5'-untranslated sequence of mRNA for human aromatase P-450". *European Journal of Biochemistry*,213 (1): 383–9.
7. Sebastian S., Serdar E Bulun (2001) Genetics of endocrine disease: A highly complex organization of the regulatory region of the human CYP19 (aromatase) gene revealed by the Human Genome Project, J Clin Endocr Metab, 86(10): 4600-4602.
8. Yang, F., *et al* (2015) Follicular hyperandrogenism downregulates aromatase in luteinizedgranulosa cells in polycystic ovary syndrome women.*Reproduction,*150(4): 289-96.
9. Rotterdam ESHRE/ASRM-Sponsored PCOS consensus workshop group (2004). Revised 2003 consensus on diagnostic criteria and long-term health risks related to polycystic ovary syndrome (PCOS). *Hum Reprod*, 19(1):41-7.
10. Franks S., Stark J and K. Hardy (2008) 'Follicle dynamics and anovulation in polycystic ovary syndrome', *Human Reproduction Update,* 14(4): 367-378
11. Jin J. L., Jing Sun., Hui-Juan Ge., Yun-Xia Cao., Xiao-Ke Wu., Feng- Jing Liang., Hai-Xiang Sun., Lu Ke., Long Yi., Zhi-Wei Wu and Yong Wang (2009)Association between CYP19 gene SNP rs2414096 Polymorphismand polycystic ovary syndrome in Chinese women.*BMC Medical Genetics* , 10:139
12. Harada N., (1992) The structural and functional characterization of P-450.aromatase (estrogen synthetase).*Tanpakushitsu KakusanKoso* ,37(16):3051-9.
13. Ito Y., Fisher C.R., Conte F.A., Grumbach M.M and E.R. Simpson (1993) Molecular basis of aromatase deficiency in an adult female with sexual infantilism and polycystic ovaries. *Proc NatlAcad Sci* USA 90: 11673-11677.
14. Belgorosky A., Pepe C, Marino R., Guercio G., Saraco N., Vaiani E and M.A. Rivarola (2003) Hypothalamic-Pituitary-ovarian axis during infancy, early and late prepuberty in an aromatase-deficient girl who is a compound heterozygote for two new point mutations of the CYP19 gene. *J Clin Endocrinol Metab* 88: 5127-5131.
15. Takayama K., Fukaya T., Sasano H., Funayama Y., Suzuki T., Takaya R., Wada Y and A. Yajima(1996)Immunohistochemical study of steroidogenesisand cell proliferation in polycystic ovarian syndrome.*Hum Reprod* , 11(7):1387-92.
16. Petry C.J, Ong K.K, Michelmore K.F, Artigas S, Wingate D.L, Balen A.H, Zegher F.D, Ibanez L and D.B.Dunger (2005) Association of *aromatase* (CYP 19) gene variation with features of hyperandrogenism in two populations of young women. *Human Reproduction*, 20(7): 1837-1843.