



# The Genome of Polycystic Ovary

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## Abstract

Polycystic ovary syndrome (PCOS) is a complex disease whose clinical manifestations and definition are an ongoing controversy among researchers. PCOS is the most common endocrinopathy in females in the childbearing period resulting in reproductive and metabolic disorders. A large number of genomic variants have been associated with PCOS, no single gene has emerged as a convincing biomarker thus far. This may be attributed to a large amount of heterogeneity observed in this disorder. This review presents an overview of the polymorphisms in genes related to insulin signaling and their association with PCOS and its related traits.

**Keywords:** Genome; PCOS; Ovary; Gene; A Metabolic Disorder

## Introduction

The polycystic ovary syndrome (PCOS) is a complex and heterogeneous endocrine condition associated with hyperandrogenism, hyperinsulinemia, insulin resistance, and chronic anovulation. Regulation and interaction of multiple genes needed for follicular development are found to be changed in PCOS Hussain [1]. The ovary is a key organ in the female genital system and its malfunction due to endocrine disturbance could lead to female infertility. Polycystic ovarian syndrome (PCOS) is one of the most common hormonal and metabolic abnormalities in females in their reproductive period Azziz [2].

## Genetic Investigations

Genetic investigations of PCOS may be divided into chromosomal/human leukocyte antigen (HLA) analysis, direct sequencing of gene regions. Karyotypes were the first genetic tests applied to study PCOS. There have been isolated case reports or small series reporting polyploidies Rojanasakul [3]. Multiple genetic causes of adult-onset hyperandrogenism and chronic anovulation have been reported. The prevalence of many of these mutations among hyperandrogenic women is still being established, although they tend to be rare compared to the relative frequency of PCOS. Mutations in steroidogenic enzymes such as 21-hydroxylase gene and 3 $\beta$ -hydroxysteroid reductase gene have been found Rheaume [4].

## Insulin Resistance and PCOS

Women suffering from PCOS are at high risk for developing diabetes. Despite the signs and symptoms of PCOS begin before the signs and symptoms of insulin resistance, insulin resistance is considered to play a role in the development of PCOS. High insulin levels may be a cofactor to inflammation and other metabolic complications described in PCOS. While the connection is thought to be known, the causes of the relationship between the two conditions are still unclear. Insulin resistance does not affect everyone in the same way, and some women with insulin resistance get PCOS, while others do not. Some authors suggest that obesity associated with insulin resistance changes the function of the hypothalamus and the pituitary gland, increasing androgenic hormones secretion, resulting in PCOS. Excessive androgenic hormones production is a risk factor for female infertility and ovarian dysfunction, with or without PCOS Nicole Galan [5].

## New Avenues in PCOS Research

Genetic studies could explore underlying genes and pathways, providing a clue of PCOS development. The results of candidate gene studies have been inconclusive, in large part due to weak studies, lack of replication and limited prior understanding of its pathogenesis Barber and Franks [6]. Mendelian randomization analyses indicate causal effects on PCOS etiology for higher body

mass index (BMI), higher insulin resistance and lower circulating serum sex hormone binding globulin (SHBG) concentrations. Furthermore, the multiple allele scores for menopausal age is positively associated with PCOS, indicating a common biological mechanism that promotes both PCOS susceptibility and then menopause Day [7].

## Conclusion

PCOS is a multifaceted disorder whose consequences extend beyond the reproductive axis and which has a major effect throughout life on the reproductive, metabolic, and cardiovascular health of affected women. The exact etiology of this multigenic and multifactorial disorder remains elusive even today despite rigorous efforts. This review has summarized the role of putative genetic variants contributing to the insulin resistance state frequently observed in PCOS women. Several pathways interlinking metabolic and reproductive processes have been dissected by studies aimed at understanding the genetic origin of this disorder.



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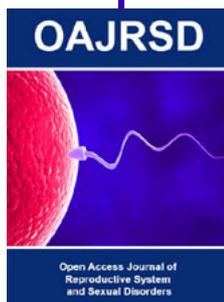
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