OPEN ACCESS Freely available online

ISSN: 2090-7214
Clinics in Mother and Child Health

Opinion Article

Impact of Genomic Factors on Androgen Levels in Polycystic Ovary Syndrome

Jingwen Ding*

Department of Obstetrics and Gynecology, Chinese Academy of Medical Sciences & Peking Union Medical College, Beijing, China

DESCRIPTION

Polycystic Ovary Syndrome (PCOS) is a common endocrine disorder that affects individuals with ovaries, primarily during their reproductive years. PCOS is characterized by a range of symptoms, including irregular menstrual cycles, ovarian cysts, and elevated levels of androgens (male sex hormones) such as testosterone. Hyperandrogenemia, the presence of excessive androgens in the bloodstream, is a key feature of PCOS and plays a central role in its pathophysiology. While the exact cause of PCOS remains elusive, recent research has discovered a genetic component associated with hyperandrogenemia in PCOS. This article explores the genetic variants linked to hyperandrogenemia in PCOS pathophysiology, providing insights on the complex interplay between genetics and hormone regulation in this prevalent condition.

Before delving into the genetic aspects of hyperandrogenemia in PCOS, it is essential to understand the clinical manifestations of this syndrome. PCOS is a heterogeneous condition, meaning that it can present differently in affected individuals. Common symptoms and signs include menstrual irregularities, hyperandrogenism, ovulatory dysfunction, insulin resistance, metabolic disturbances, and psychological impact.

PCOS often manifests as irregular or absent menstrual periods, making it challenging for affected individuals to predict ovulation or plan for pregnancy. Elevated levels of androgens can lead to symptoms such as hirsutism (excessive hair growth), acne, and male-pattern baldness. PCOS can disrupt the normal ovulation process, leading to fertility issues and an increased risk of ovarian cysts. Many individuals with PCOS also experience insulin resistance, which can contribute to weight gain and an increased risk of developing type-2 diabetes. PCOS is associated with metabolic abnormalities, including dyslipidemia and an increased risk of cardiovascular disease. The physical symptoms and fertility challenges associated with PCOS can have a negative impact on mental health, leading to depression and anxiety in some cases.

While PCOS is considered a complex genetic disorder, it is also influenced by environmental factors. Genetic studies have shown that PCOS has a hereditary component, meaning that it can run in families. However, PCOS does not follow a straightforward Mendelian inheritance pattern, where a single gene mutation leads to the condition. Instead, PCOS is thought to involve multiple genetic variants, each contributing to an individual's risk.

Researchers have made significant strides in identifying genetic variants associated with PCOS, particularly those related to hyperandrogenemia. Several genes have been implicated in the regulation of androgen levels in individuals with PCOS. Some of the key genetic variants associated with hyperandrogenemia in PCOS include *INSR* (Insulin Receptor Gene), *LHCGR* (Luteinizing Hormone/Choriogonadotropin Receptor Gene), *CYP17A1* (Cytochrome P450 17A1), *DENND1A* (DENN Domain-Containing Protein 1A), *FSTL3* (Follistatin-like 3), *AR* (Androgen Receptor Gene).

Insulin resistance is a common feature of PCOS, and variations in the *INSR* gene have been linked to insulin resistance, which, in turn, can contribute to hyperandrogenemia. Variants in the *LHCGR* gene can lead to increased responsiveness of ovarian cells to Luteinizing Hormone (LH), resulting in elevated androgen production. This gene encodes an enzyme essential for androgen synthesis. Variants in *CYP17A1* can influence the enzyme's activity, leading to increased androgen production. *DENND1A* is involved in the regulation of androgen biosynthesis in ovarian theca cells. Genetic variants in this gene may contribute to hyperandrogenemia in PCOS. *FSTL3* is involved in the regulation of the Gonadotropin-Releasing Hormone (GnRH) pulse generator. Variants in this gene may affect GnRH secretion and subsequently impact androgen levels. Variations in the androgen receptor gene can influence androgen sensitivity, leading to increased androgenic effects even with normal androgen levels.

The genetic variants associated with hyperandrogenemia in PCOS are not isolated factors but rather components of a complex regulatory network. The pathophysiology of PCOS involves interactions between multiple genes, hormonal pathways, and environmental factors. Genetic variants in *INSR*

Correspondence to: Jingwen Ding, Department of Obstetrics and Gynecology, Chinese Academy of Medical Sciences & Peking Union Medical College, Beijing, China, E-mail: jingwending@foxmail.com

Received: 22-Aug-2023, Manuscript No. CMCH-23-23469; **Editor assigned:** 25- Aug-2023, Pre QC No. CMCH-23-23469 (PQ); **Reviewed:** 08-Sep-2023, QC No. CMCH-23-23469; **Revised:** 15-Sep-2023, Manuscript No. CMCH-23-23469 (R); **Published**: 22-Sep-2023, DOI: 10.35248/2090-7214.23.S18.004

Citation: Ding J (2023) Impact of Genomic Factors on Androgen Levels in Polycystic Ovary Syndrome. Clinics Mother Child Health. S18:004.

can lead to insulin resistance, which in turn stimulates the ovaries to produce more androgens. This insulin resistance exacerbates hyperandrogenemia. Genetic variations in *LHCGR* can result in ovarian cells being more sensitive to LH, a hormone that stimulates androgen production in the ovaries. This heightened sensitivity contributes to elevated androgen levels. Variants in genes like *CYP17A1* and *DENND1A* can directly affect androgen synthesis, increasing the production of these hormones in the ovaries. *FSTL3* variants may influence the pulsatile secretion of GnRH, a hormone that controls the release of LH and Follicle-Stimulating Hormone (FSH). Dysregulation of GnRH secretion can disrupt the ovarian hormonal balance, leading to hyperandrogenemia. Genetic variations in the Androgen Receptor Gene (*AR*) can make tissues more sensitive to androgens, even at normal circulating levels, intensifying androgenic effects.

Understanding the genetic variants associated with hyperandrogenemia in PCOS is a significant step toward personalized medicine and targeted therapies. It allows healthcare providers to identify individuals at higher risk for severe androgen-related symptoms and tailor treatments accordingly. For example, medications that target specific pathways affected by these genetic variants may be more effective in managing hyperandrogenemia in PCOS. Future research in this field will likely uncover additional genetic factors and provide insights into the precise mechanisms underlying PCOS pathophysiology. Moreover, ongoing studies may help identify novel therapeutic targets and strategies aimed at mitigating the impact of genetic variants on androgen levels in PCOS.

CONCLUSION

Polycystic Ovary Syndrome is a multifaceted condition with a complex genetic underpinning. The presence of hyperandrogenemia in PCOS is influenced by various genetic variants that impact hormonal pathways and androgen production. Understanding these genetic factors is essential for developing more targeted and effective treatments for PCOS, ultimately improving the quality of life for individuals affected by this syndrome. Even though there has been a lot to to discover in this field, current research offers expectations for more personalized and precise interventions for PCOS-related hyperandrogenemia.