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Polycystic ovary syndrome (PCOS): A Detailed Review of Pathophysiology, clinical Manifestations, Diagnosis and management.

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Abstract

Polycystic Ovary Syndrome (PCOS) is one of the most common endocrine and metabolic disorders affecting women of reproductive age. It is characterized by hyperandrogenism, ovulatory dysfunction, and polycystic ovarian morphology.(1) The disorder has complex and multifactorial origins involving genetic, hormonal, metabolic, and environmental factors. PCOS significantly affects reproductive, metabolic, cardiovascular, and psychological health.(2) Women with PCOS frequently experience menstrual irregularities, infertility, obesity, insulin resistance, and psychological disturbances. Early diagnosis and individualized treatment approaches are essential to improve quality of life and prevent long-term complications. This review discusses epidemiology, etiology, pathophysiology, clinical manifestations, diagnostic criteria, complications, and current management strategies of PCOS.

Keywords:
Polycystic Ovary Syndrome, Hyperandrogenism, Insulin Resistence, Infertility, Metabolic Syndrome, Harmonal Disorders

Introduction

Polycystic Ovary Syndrome (PCOS), previously called Stein-Leventhal syndrome, is a heterogeneous endocrine disorder affecting women during reproductive years. It represents one of the leading causes of anovulatory infertility worldwide. The prevalence varies depending on diagnostic criteria but generally ranges from approximately 6-20%.

PCOS affects reproductive and metabolic systems simultaneously. The syndrome presents with a broad spectrum of symptoms, including menstrual irregularities, obesity, hirsutism, acne, infertility, and metabolic abnormalities. Despite extensive research, the exact etiology remains unclear because multiple mechanisme interact in disease development.

Epidemiology

PCOS prevalence differs according to population characteristics and diagnostic methods.

Factors affecting prevalence include:

- Ethnicity
- Genetic susceptibility
- Lifestyle patterns
- Obesity prevalence

Diagnostic criteria used

Studies suggest that obesity increases disease severity and metabolic complications.

Etiology

The etiology of PCOS is multifactorial and includes:

Genetic Factors

Family studies indicate strong hereditary influences. Multiple genes involved in insulin signaling and androgen synthesis contribute to susceptibility.

Genes potentially associated include:

- CYP11A
- FSH receptor genes
- Insulin receptor genes
- Androgen receptor genes

Environmental Factors

Environmental influences include:

- High-calorie diets
- Sedentary lifestyle
- Obesity
- Exposure to endocrine-disrupting chemicals
- Stress

Hormonal Factors

Hormonal abnormalities include:

- Increased luteinizing hormone secretion
- Increased androgen production
- Disturbed gonadotropin release

- Elevated Anti-Müllerian Hormone (AMH)

Pathophysiology

The pathophysiology of PCOS involves interactions among insulin resistance, ovarian dysfunction, and hormonal imbalance.

Insulin Resistance

Insulin resistance is present in many women with PCOS and is considered a major pathological mechanism.

Consequences include:

- Increased insulin secretion
- Hyperinsulinemia
- Increased ovarian androgen production
- Reduced sex hormone-binding globulin (SHBG)

Reduced SHBG increases circulating free testosterone.

Hyperandrogenism

Hyperandrogenism results in:

- Hirsutism
- Acne
- Alopecia
- Menstrual disturbances

Excess androgen production interferes with normal follicular development.

Hypothalamic-Pituitary-Ovarian Axis Dysfunction

Abnormal secretion of gonadotropin-releasing hormone leads to:

- Increased LH secretion

- Reduced FSH secretion
- Elevated LH ratio

This imbalance disrupts ovulation.

Follicular Arrest

Multiple immature follicles accumulate in ovaries due to failure of dominant follicle selection.

Consequences include:

- Chronic anovulation
- Infertility
- Polycystic ovarian appearance

Clinical Manifestations

Clinical manifestations vary among patients.

Reproductive Symptoms

- Irregular menstrual cycles
- Oligomenorrhea
- Amenorrhea
- Infertility
- Recurrent miscarriage

Hyperandrogenic Symptoms

- Hirsutism
- Acne vulgaris
- Male-pattern hair loss
- Seborrhea

Metabolic Features

- Weight gain
- Central obesity
- Dyslipidemia
- Insulin resistance

Dermatological Features

- Acanthosis nigricans
- Skin tags
- Oily skin

Psychological Manifestations

- Depression
- Anxiety
- Mood disorder
- Reduced self-esteem
- Sleep disturbances

Diagnosis

There is no single diagnostic test for PCOS.

The Rotterdam criteria require at least two of the following:

1. Oligo-ovulation or anovulation
2. Clinical or biochemical hyperandrogenism
3. Polycystic ovaries on ultrasound

Laboratory Investigations

- Serum testosterone
- LH and FSH
- Prolactin
- Thyroid function tests
- Fasting glucose
- Insulin levels
- Lipid profile

Imaging Studies

Pelvic ultrasonography demonstrates:

- Enlarged ovaries
- Multiple small follicles
- Peripheral arrangement of follicles

Complications

Long-term complications include:

Metabolic Complications

- Type 2 diabetes mellitus
- Metabolic syndrome
- Dyslipidemia
- Hypertension

Reproductive Complications

- Infertility
- Pregnancy complications
- Endometrial hyperplasia
- Endometrial cancer

Cardiovascular comlications

- Increased cardiovascular risk
- Endothelial dysfunction

Psychological Complications

- Depression
- Anxiety disorders
- Eating disorders

Management

Management should be individualized according to symptoms and fertility goals.

Lifestyle Modification

First-line treatment includes:

- Weight reduction
- Regular exercise
- Dietary modifications
- Behavioral interventions

Benefits include:

- Improved insulin sensitivity
- Better menstrual regulation
- Reduced androgen levels

Pharmacological Treatment

Oral Contraceptive Pills

Used to:

- Regulate menstrual cycles
- Reduce androgen effects
- Improve acne

Metformin

Benefits include:

- Increased insulin sensitivity

- Improved Ovulation
- Reduction in glucose levels

Anti-androgen Therapy

Examples :

- Spironolactone
- Finasteride

Used for:

- Hirsutism
- Acne

Fertility Treatment

Options include :

- Letrozole
- Clomiphene citrate
- Gonadotropins
- Assisted reproductive technologies

Emerging Therapies

Recent developments include:

- Inositol supplementation
- GLP-1 receptor agonists
- Gut microbiota interventions
- Personalized medicine approaches
- Artificial intelligence-based diagnostic tools

Future Perspectives

Future research should focus on:

- Identification of genetic markers
- Precision medicine
- Improved diagnostic biomarkers
- Novel targeted therapies.

Conclusion

PCOS is a multifactorial endocrine disorder affecting reproductive, metabolic, and psychological health. Early diagnosis and individualized treatment strategies are essential to minimize complications and improve quality of life. Lifestyle modification remains the cornerstone of therapy, while emerging treatments may further improve long-term outcomes

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