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Association of Apolipoprotein E Gene Polymorphisms with Polycystic Ovary Syndrome: A Case-Control Study

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ABSTRACT

Objective: To compare the distribution of ApoE polymorphisms between women with PCOS and healthy controls to explore whether specific ApoE genotypes contribute to the genetic susceptibility of developing PCOS, and to investigate the association between APOE gene polymorphisms and lipid profiles in PCOS patients.

Methods: A case-control study was conducted between November 2023 and January 2025, enrolling 120 women with PCOS diagnosed according to Rotterdam criteria and 60 age-matched healthy controls. Participants underwent comprehensive clinical assessment, hormonal evaluation (FSH, LH, total and free testosterone), lipid profiling, and inflammatory marker analysis. DNA extraction was performed from whole blood, followed by PCR amplification and direct sequencing of ApoE gene fragments containing SNPs rs429358 and rs7412.

Results: PCOS participants demonstrated significantly higher age, body weight, and height compared to controls ($p<0.05$). Hormonal analysis revealed characteristic PCOS patterns with elevated LH, total testosterone, free testosterone, and C-reactive protein levels, alongside reduced FSH concentrations ($p<0.001$). Lipid profile analysis showed significantly higher total cholesterol and LDL levels, with lower HDL concentrations in PCOS patients ($p<0.05$). Genetic analysis identified three ApoE genotypes (ε3ε3, ε2ε3, ε3ε4), with ε3ε3 being most prevalent in both groups. No significant differences were observed in ApoE genotype or allele distribution between PCOS patients and controls ($p>0.05$). However, within the PCOS group, ε4 allele carriers exhibited significantly elevated total cholesterol ($p=0.039$), triglycerides ($p=0.013$), and VLDL levels ($p=0.026$) compared to ε2 and ε3 carriers.

Conclusions: ApoE gene polymorphisms do not appear to significantly influence PCOS susceptibility, as genotype distributions were comparable between patients and controls. However, ApoE variants, particularly the ε4 allele, may modulate metabolic dysfunction severity in women with established PCOS, potentially affecting cardiovascular risk stratification and therapeutic management approaches.

Keywords: Apolipoprotein E, Gene polymorphisms, Lipid metabolism, Polycystic Ovary Syndrome.

INTRODUCTION

Polycystic Ovary Syndrome (PCOS) is one of the most prevalent endocrine disorders affecting women of reproductive age, with an estimated prevalence ranging from 5% to 20% depending on the diagnostic criteria used [(1)].

This complex multisystem disorder is characterized by a constellation of symptoms including hyperandrogenism, ovulatory dysfunction, and polycystic ovarian morphology, often accompanied by significant metabolic disturbances [(2)]. Beyond its reproductive manifestations, PCOS is frequently associated with insulin resistance, dyslipidemia, obesity, and an increased risk of cardiovascular disease and type 2 diabetes mellitus, making it a significant public health concern [(3)].

The etiology of PCOS remains incompletely understood, though it is widely recognized as a multifactorial disorder involving complex interactions between genetic predisposition and environmental factors [(4)]. The strong familial clustering observed in PCOS suggests a substantial genetic component, with heritability estimates ranging from 65% to 70% [(5)]. Identifying specific genetic markers that contribute to PCOS susceptibility is crucial for understanding its pathogenesis, improving diagnostic accuracy, and developing targeted therapeutic interventions.

Apolipoprotein E (ApoE) is a critical lipid-binding protein that plays a central role in lipid metabolism and transport, particularly in the clearance of triglyceride-rich lipoproteins and cholesterol homeostasis [(6)]. The APOE gene exhibits three major polymorphic variants (ε2, ε3, and ε4), which encode three distinct protein isoforms (ApoE2, ApoE3, and ApoE4) with different functional properties [(7)]. These polymorphisms have been extensively studied concerning cardiovascular disease, with the ε4 allele associated with increased cardiovascular risk and the ε2 allele generally considered protective [(8)].

Given the prominent metabolic disturbances inherent in PCOS, particularly the dyslipidemia and increased cardiovascular risk, investigating the potential association between APOE gene polymorphisms and PCOS susceptibility represents a logical area of inquiry. Previous studies have yielded conflicting results regarding this association, with some suggesting potential links between specific ApoE genotypes and PCOS risk or metabolic phenotypes, while others have found no significant associations [(9,10)]. This study aims to compare the distribution of ApoE polymorphisms between women with PCOS and healthy controls to explore whether specific ApoE genotypes contribute to the genetic susceptibility of developing PCOS

MATERIALS AND METHODS

A case-control investigation was carried out between November 2023 and January 2025. The research enrolled 120 women diagnosed with polycystic ovary syndrome (PCOS) based on the Rotterdam criteria established in 2003 [(11)], confirmed by a board-certified gynecologist. Ethical approval for this research was obtained from the Institutional Review Board at Al-Nahrain University's College of Medicine (Reference Number: 20231013, dated January 18, 2024). Written informed consent was secured from all study participants.

The inclusion criteria encompassed women with PCOS aged 20-50 years having a body mass index below 35 kg/m2. Exclusion criteria comprised participants presenting with hypertension, coronary artery disease, cardiac valve disorders, congestive heart failure, pyrexia, menopausal status, rheumatoid arthritis, autoimmune conditions, persistent infections, or a BMI equal to or exceeding 35 kg/m2. Additionally, 60 age-matched healthy women were recruited to serve as the control group (HCs).

Clinical Assessment and Physical Evaluation

The assessment protocol encompassed a structured questionnaire covering participant demographics including age, tobacco use status, educational background, and socioeconomic status, along with anthropometric measurements comprising weight, height, body mass index (BMI), and waist-to-hip ratio (WHR).

Biochemical Assessments

Hormonal parameters including follicle-stimulating hormone (FSH), luteinizing hormone (LH), and total testosterone concentrations were determined utilizing the Cobas e411 analyzer (Roche, Germany). Free testosterone levels were quantified through enzyme-linked immunosorbent assay (ELISA) methodology using Kayto kits (China). The comprehensive lipid panel encompassing total cholesterol (TC), triglycerides (TG), high-density lipoprotein cholesterol (HDL), low-density lipoprotein cholesterol (LDL), and very low-density lipoprotein cholesterol (VLDL) was assessed employing the Cobas c311 system (Roche, Germany). Additionally, C-reactive protein (CRP) concentrations were measured using the same Cobas c311 platform (Roche, Germany).

DNA extraction, Gene amplification and genotyping [(12)]

Genomic DNA isolation from whole blood samples was performed utilizing a commercially available extraction kit (gSYNCTM DNA Mini Kit Whole Blood Protocol/ Genaid/ Korea) according to the provided manufacturer's protocol. PCR amplification of the 720 base pair ApoE gene segments containing the polymorphic sites Rs429358 and Rs7412 was conducted using designated oligonucleotide primer pairs (Bioneer, Korea). The primer sequences employed in this study were as detailed below: Fwd: 5'-GGACGAGACCATGAAGGAGTT-3` and Rev: 5'-GCTTCGGCGTTCAGTGATTGT-3`. The polymerase chain reaction was performed in a 25µl reaction mixture comprising 50 ng genomic DNA template, 1.5 µl 10×PCR buffer, 0.3 µl 10 mM dNTPs, 0.25 µl 10 pmol/µl of

each oligonucleotide primer, and 1.25 U Taq DNA polymerase (Bioneer/Korea). Thermal cycling was conducted using an ABI 9600 thermocycler (Hybaid, England) under the following parameters: initial denaturation at 95°C for 4 min, followed by 35 cycles of denaturation at 94°C for 45 s, annealing at 61°C for 30s, and extension at 72°C for 45 s, with a final extension phase at 72°C for 7 min. Following amplification, the PCR products were gel-purified and submitted for DNA sequencing services (Macrogen, Korea). Sequence verification and analysis were performed through comparative alignment using the Basic Local Alignment Search Tool (BLAST) platform accessible at https://blast.ncbi.nlm.nih.gov/Blast.cgi.

Statistical analysis

Statistical analyses were performed by using SPSS software version 25.0 (SPSS, Chicago). Continuous data were presented as mean and standard deviation and analyzed with Student t-test. Categorical variables were expressed as number and percentage and analyzed with the Chi-square test. Binary logistic regression was used to determine the association between APOE gene polymorphism and PCOS. The odds ratio (OR) and its corresponding 95% confidence interval (CI) were derived from this test. A p- value less than 0.05 was considered to indicate a statistically significant difference.

RESULTS

Participants with PCOS demonstrated significantly advanced age (p= 0.002) and exhibited greater body weight and stature (p <0.001, respectively) when compared to healthy controls (HCs). Educational attainment at the university level and beyond was less prevalent among PCOS participants compared to the control group (p= 0.004). Conversely, no statistically significant differences were observed between the groups regarding body mass index, waist-to-hip ratio, tobacco use history, or monthly earnings.

Table 1: Demographic Characteristics and Baseline Population Data (n±SD, p-value < 0.05)

Variables	Patients (n=120)	Controls (n=60)	p-value
Age, years			
Mean±SD	32.98±7.28	29.62±5.9	**0.002**
Range	20-48	20-47	
Weight, kg			
Mean±SD	70.61±9.49	64.13±8.31	**<0.001**
Range	51-91	49-84	
Height, m			
Mean±SD	1.6±0.07	1.53±0.06	**<0.001**
Range	1.44-1.75	1.43-1.67	
Body mass index, kg/m^2			
Mean±SD	27.64±3.51	27.25±3.02	0.470
Range	20.9-34.8	21.4-35.0	
Waist-hip ratio			
Mean±SD	0.82±0.1	0.84±0.06	0.089
Range	0.65-1.05	0.72-0.97	
Smoking history			
Never	111 (92.5%)	57(95%)	0.388
Ex/current	9(7.5%)	3(5%)	
Education level			
Primary school	3(2.5%)	1(1.67%)	**0.004**
Intermediate school	46(38.33%)	11(18.33%)	
Secondary school	49(40.83%)	23(38.33%)	
University	18(15%)	16(26.67%)	
Higher	4(3.33%)	9(15%)	
Income/month			
Low	45(37.5%)	23(38.33%)	
Moderate	61(50.83%)	25(41.67%)	0.268
High	14(11.67%)	12(20%)	

Endocrine Parameters and Inflammatory Markers

Table 2 demonstrates that participants with PCOS displayed significantly reduced follicle-stimulating hormone (FSH) concentrations alongside elevated luteinizing hormone (LH), free testosterone, total testosterone, and C-reactive protein levels in serum ($p < 0.001$) compared to healthy controls.

Table 2: Endocrine profile and inflammatory biomarkers in the study cohort (n±SD, p-value < 0.05)

Variables	Patients (n=120)	Controls (n=60)	p- value
FSH, mIU/ml			
Mean ±SD	6.49±0.67	8.34±2.38	**<0.001**
Range	4.17-8.24	4.02-12.29	
LH, mIU/ml			
Mean ±SD	13.55±0.78	8.08±2.25	**<0.001**
Range	11.36-16.21	3.89-11.80	
Total Testosterone, ng/ml			
Mean ±SD	0.92±0.28	0.34±0.11	**<0.001**
Range	0.47-1.90	0.09-0.54	
Free Testosterone, ng/ml			
Mean ±SD	2.95±0.47	1.3±0.4	**<0.001**
Range	2.10-4.72	0.17-1.90	
C-reactive protein, mg/L			
Mean ±SD	3.09±1.05	2.2±1.2	**<0.001**
Range	1.1-4.8	0.5-4.6	

Lipid Profile

Table 3 shows higher levels of TC and LDL were demonstrated in PCOS women compared to HCs ($p= 0.009$; $p< 0.001$, respectively). Conversely, HDL levels were significantly lower in the PCOS group than in the HCs ($p< 0.001$). Although TG and VLDL levels were elevated in PCOS patients, these differences did not reach statistical significance.

Table 3: Lipid profile of the study population (n±SD, p-value < 0.05)

Variables	Patients (n=120)	Controls (n=60)	p- value
Cholesterol, mg/dL			
Mean ±SD	182.49±24.09	172.84±13.28	**0.009**
Range	139.9-255.8	142.8-204.6	
TG, mg/dL			
Mean ±SD	125.29±28.14	119.78±22.64	0.228
Range	91.1-211.5	79.6-161.7	
HDL, mg/dL			
Mean ±SD	35.96±4.42	40.30±3.87	**<0.001**
Range	24.40-44.3	30.1-47.2	
LDL, mg/dL			
Mean ±SD	121.13±19.06	107.20±7.53	**<0.001**
Range	82.8-180.2	87.4-126.6	
VLDL, mg/dL			
Mean ±SD	24.56±6.22	23.78±4.40	0.435
Range	12.9-42.3	15.9-31.9	

Molecular Assays

To investigate the association between ApoE genetic variants and PCOS, we analyzed two specific single nucleotide polymorphisms using standard PCR amplification techniques with targeted primer sequences. The electrophoretic separation of the amplified ApoE gene segment, which encompasses the polymorphic sites rs429358 and rs7412, is illustrated in Figure 1. Direct DNA sequencing was employed to determine the genotypic profiles of the study participants

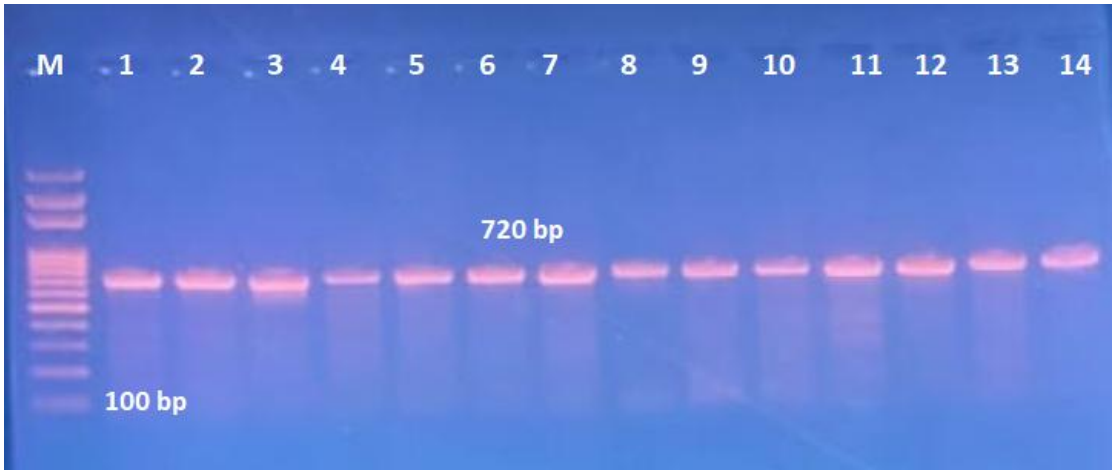

Figure 1. Electrophoretic separation of the Apolipoprotein E (ApoE) gene segment (720 base pairs) containing the single nucleotide polymorphisms rs429358 and rs7412.

Sequencing of SNP rs429358 showed two genotypes (TT and CT) in patients and controls (Figure 2).

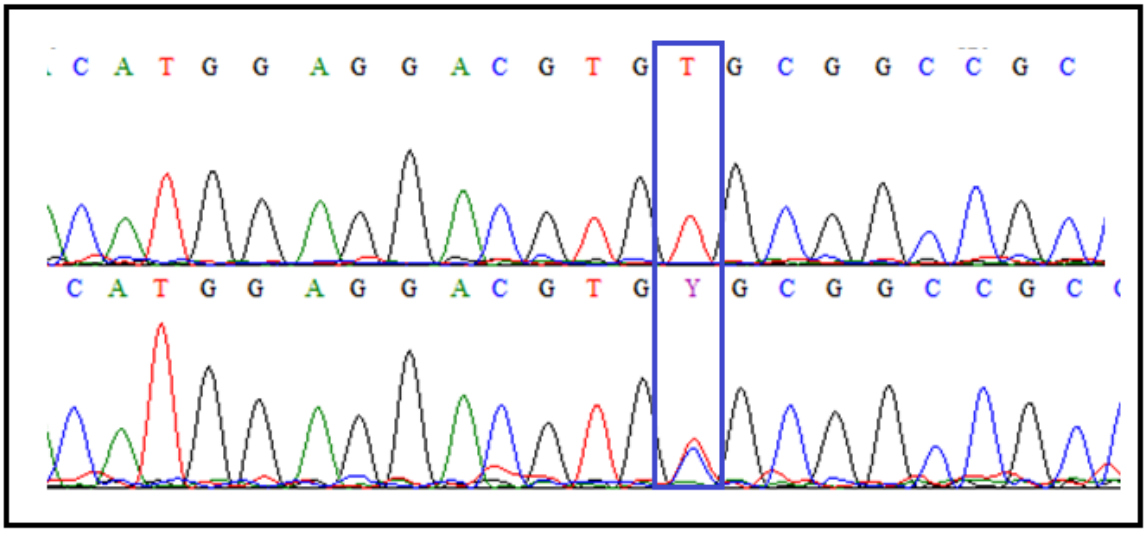

Figure 2. Sequencing chromatogram showing the forward strand of SNP rs429358. The highlighted regions indicate the polymorphic sites. The upper frame displays a T nucleotide, representing the homozygous wild type genotype (TT), while the lower frame shows a Y symbol (indicating a C/T nucleotide mixture), representing the heterozygous genotype (CT).

The frequency of the heterozygous genotype (CT) genotype was higher in PCOS than controls (17.5% vs. 10%); however, the difference was not significant. Similarly, C allele was slightly higher in patients than controls (8.7% vs. 5%) with no significant difference (Table 4).

Table 4: Distribution of rs429358 Polymorphism Genotypes and Allele Variants Within the Study Cohort (p-value < 0.05)

Rs429358 Polymorphism	Patients (120)	Controls (60)	*P*-value	OR(95%CI)
Genotypes				
TT	99(82.5%)	54(90%)	0.190	1.0
CT	21(17.5%)	6(10%)		1.91(0.27-5.02)
Alleles				
T	219(91.25%)	114(95%)	0.254	1.0
C	21(8.75%)	6(5%)		1.73(0.68-4.42)

While the SNP rs7412 showed in two genotypes (CC and CT) in patients and controls (Figure 3).

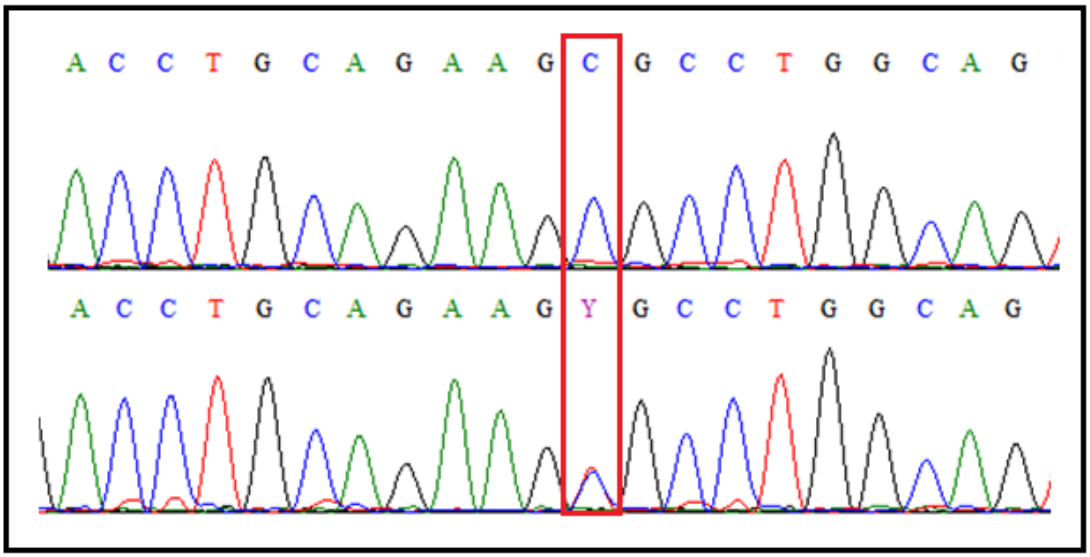

Figure 3. Sequencing chromatogram displaying the forward strand of SNP rs7412. The highlighted areas indicate the polymorphic positions. The upper frame shows a C nucleotide, representing the homozygous wild type genotype (CC), while the lower frame displays a Y symbol (indicating a C/T nucleotide mixture), representing the heterozygous genotype (CT).

Table 5 presents the genotypic and allelic frequencies of rs7412 SNP in PCOS patients and controls with no significant differences.

Table 5: Distribution of rs7412 Polymorphism Genotypes and Allele Variants Within the Study Cohort (p-value < 0.05)

Rs7412 Polymorphism	Patients (120)	Controls (60)	*P*-value	OR(95%CI)
Genotypes				
CC	107(89.17%)	53(88.33%)	0.867	1.0
CT	13(10.83%)	7(11.67%)		0.92(0.35-2.44)
Alleles				
C	227(94.58%)	113(94.17%)	0.739	1.0
T	13(5.42%)	7(11.67%)		0.85(0.33-2.22)

Classification of Genotypes Based on Various Epsilon Allele Combinations

Various epsilon genotypes are created through combinations of alleles from the first SNP (rs429358) with alleles from the second SNP (rs7412). This study identified three genotypes: ε3 ε3, ε2 ε3, and ε3 ε4. The ε3 ε3 genotype was found to be the most prevalent and was therefore designated as the wild type genotype. No significant differences were found between the groups for genotype or allele distribution, implying that epsilon genotype variation may not play a major role in PCOS susceptibility as shown in Table 6.

Table 6: The frequency of epsilon genotypes and allele in patients and controls (p-value < 0.05)

Epsilon	Patients (120)	Controls (60)	*P*-value	OR(95%CI)
Genotypes				
ε3ε3	89(74.17%)	47(78.33%)	0.730	1.0
ε2ε3	14(11.67%)	7(11.67%)	0.428	1.5(0.55-4.05)
ε3ε4	17(14.17%)	6(10%)	0.599	1.42(0.39-5.2)
Alleles				
ε3	209(87.08%)	107(89.17%)	0.749	1.0
ε2	14(5.83%)	7(5.83%)	0.447	1.45(0.56-3.79)
ε4	17(7.08%)	6(5%)	0.599	1.42(0.87-5.2)

Association of APOE gene polymorphisms with lipid profiles

PCOS individuals carrying the ε4 allele exhibited significantly elevated levels of total cholesterol (p= 0.039), triglycerides (p= 0.013), and VLDL (p= 0.026) when compared to those with ε2 and ε3 alleles (Table 7).

Table 7: Association of APOE gene polymorphisms in PCOS group with lipid profile (n±SD, p-value < 0.05)

Variables	ε3 (n=89)	ε2 (n= 14)	ε4 (n=17)	p-value
TC	179.65±23.23	184.47±23.66	195.67±25.67	**0.039**
TG	121.55±24.01	127.43±30.08	143.14±39.51	**0.013**
HDL	35.74±4.46	36.80±4.09	36.44±4.63	0.633
LDL	119.56±18.45	121.2±19.11	129.3±21.15	0.155
vLDL	23.79±5.43	25.04±6.53	28.17±8.6	**0.026**

DISCUSSION

The present investigation revealed that participants with PCOS demonstrated advanced age alongside increased body weight and stature. These findings are consistent with prior research suggesting that PCOS manifestations may intensify over time as a result of progressive metabolic and endocrine dysfunction [13,14]. While aging itself is not a causative factor for PCOS, it may amplify the syndrome's clinical presentation by exacerbating insulin resistance (IR) and androgen excess [15]. The higher average body weight observed in PCOS participants corresponds with established literature reporting elevated rates of excess weight and obesity, including among those with similar BMI values compared to their non-PCOS peers [15].

The PCOS participants in this research demonstrated reduced levels of tertiary and post-secondary educational attainment compared to healthy controls. Previous research has established connections between diminished academic achievement and decreased health awareness, resulting in prolonged delays in seeking specialist healthcare services. This pattern subsequently contributes to postponed diagnostic processes for women experiencing PCOS symptoms [16].

Regarding the hormonal characteristics, women diagnosed with PCOS characteristically demonstrate increased luteinizing hormone (LH), total and free testosterone concentrations, alongside reduced follicle-stimulating hormone (FSH) levels. Earlier investigations have repeatedly documented this endocrine pattern and attributed it to disruption of the hypothalamic-pituitary-ovarian axis function [17,18]. The elevated C-reactive protein (CRP) concentrations observed in PCOS participants within this investigation may suggest the presence of an underlying chronic subclinical inflammatory state [19].

The present investigation revealed a characteristic lipid abnormality pattern among PCOS participants, featuring raised total cholesterol (TC) concentrations, heightened low-density lipoprotein (LDL) levels, and reduced high-density lipoprotein (HDL) concentrations, while triglyceride (TG) and very low-density lipoprotein (VLDL) values showed no significant variation. These findings align with a growing body of evidence indicating that lipid disturbances are a prominent metabolic feature of PCOS [20,21].

However, not all studies concur with the extent of TG and VLDL changes. Some earlier research in lean or young PCOS populations found no significant difference in TG or TC levels compared to controls, with PCOS subjects differing mainly in having lower HDL and higher LDL [22]. Such discrepancies are often attributed to variations in sample characteristics (e.g. age, adiposity, ethnicity) and the criteria used to define PCOS.

ApoE gene polymorphisms and polycystic ovary syndrome

In this study, the distribution of ApoE genotypes for both rs429358 and rs7412 polymorphism, and their resulting ε2/ε3/ε4 isoforms, showed no statistically significant difference between PCOS patients and HCs apart from ε3/ε3 being the most common in both groups.

Following these findings, a study from China also found no significant difference in the distribution of ApoE genotypes or alleles between Chinese women with PCOS and HCs [23]. They also conclude that ApoE polymorphisms are not strongly associated with PCOS susceptibility. Another case-control study on 58 PCOS patients and 91 healthy women from Finland, also reported no link between ApoE polymorphisms and PCOS, corroborating the above findings [9]. Two cohort studies from the UK and Ireland on 137 PCOS subjects and 97 controls also found ApoE2, ApoE3, and ApoE4, were comparable between PCOS subjects and controls [24].

These consistent null findings span different ethnicities implying that the genetic isoform distribution remained unchanged, and not associated with PCOS risk, reinforcing that ApoE is probably is not a primary genetic determinant of PCOS.

Despite the overall negative evidence, a few studies have reported significant ApoE frequency differences in PCOS, suggesting a potential association. A Turkish study on 129 PCOS women and 91 HCs found that ApoE

allele distributions in PCOS patients deviated from controls. The ApoE3 allele was more frequent in PCOS patients, whereas the ApoE2 allele was over-represented in HCs [25]. Specifically, the wild-type ε3 allele appeared to confer a higher risk, and the ε2 allele might have had a protective association in that population.

Associations of ApoE polymorphisms with lipid profile

The analysis of APOE genotypes in women with PCOS revealed notable associations with lipid profile, specifically, ε4 allele carriers exhibited significantly higher levels of TC, TG, and VLDL compared to ε2 and ε3 carriers.

The observation of higher TC and LDL in ε4 carriers is in line with countless reports in general populations [26, 27, 28].

CONCLUSION

This case-control study investigated the potential association between ApoE gene polymorphisms and PCOS susceptibility in women of reproductive age. Our findings demonstrate that the distribution of ApoE genotypes (rs429358 and rs7412) and their resulting ε2/ε3/ε4 isoforms did not differ significantly between PCOS patients and healthy controls, suggesting that ApoE polymorphisms are not primary genetic determinants of PCOS susceptibility. These results align with previous studies from diverse ethnic populations, reinforcing the notion that ApoE variants do not substantially contribute to PCOS risk. However, our analysis revealed clinically relevant associations between ApoE genotypes and metabolic profiles within the PCOS population, particularly among ε4 allele carriers who exhibited significantly elevated levels of total cholesterol, triglycerides, and VLDL compared to ε2 and ε3 carriers. This finding suggests that while ApoE polymorphisms may not influence PCOS development, they could modulate the severity of metabolic dysfunction in affected women. These insights have important implications for personalized risk stratification and therapeutic management of PCOS patients, particularly regarding cardiovascular risk assessment. Future research should focus on larger multicenter studies and explore the interaction between ApoE variants and other genetic factors in determining PCOS phenotypic heterogeneity and long-term metabolic outcomes.

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CONFLICTS OF INTEREST

The authors declare no conflicts of interest regarding the publication of this manuscript.

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ETHICS STATEMENTS

This study was conducted following the Declaration of Helsinki and was approved by the Institutional Review Board/Ethics Committee of Al-Nahrain University's College of Medicine (Approval No. 20231013, dated 18/1/2024). All procedures performed in this study involving human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

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دراسة الارتباط بين تعدد الأشكال الجينية لبروتين أبوليبوبروتين إي ومتلازمة المبيض متعدد الكيسات: دراسة تحليلية للحالات والضوابط

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الخلاصة

الهدف: مقارنة توزيع تعدد الأشكال الجينية لبروتين أبوليبوبروتين إي (ApoE) بين النساء المصابات بمتلازمة المبيض متعدد الكيسات والضوابط الصحية لاستكشاف ما إذا كانت الأنماط الجينية المحددة لـ ApoE تساهم في القابلية الوراثية لتطوير متلازمة المبيض متعدد الكيسات، والتحقق من الارتباط بين تعدد الأشكال الجينية لجين APOE وملفات الدهون في مريضات متلازمة المبيض متعدد الكيسات. **طرق العمل:** أُجريت دراسة حالات وضوابط بين نوفمبر 2023 ويناير 2025، شملت 120 امرأة مصابة بمتلازمة المبيض متعدد الكيسات مشخصة وفقاً لمعايير روتردام و60 ضابطة صحية متطابقة في العمر. خضعت المشاركات لتقييم سريري شامل، وتقييم هرموني (الهرمون المنشط للجريب، الهرمون اللوتيني، التستوستيرون الكلي والحر)، وتحليل ملف الدهون، وتحليل علامات الالتهاب. تم استخلاص الحمض النووي من الدم الكامل، تبعه تضخيم تفاعل البوليميراز المتسلسل والتسلسل المباشر لأجزاء جين ApoE المحتوية على تعدد النوكليوتيدات المفردة rs429358 و rs741 . **النتائج:** أظهرت مشاركات متلازمة المبيض متعدد الكيسات عمراً ووزن جسم وطولاً أعلى بشكل كبير مقارنة بالضوابط .($p<0.05$) كشف التحليل الهرموني عن أنماط مميزة لمتلازمة المبيض متعدد الكيسات مع ارتفاع مستويات الهرمون اللوتيني والتستوستيرون الكلي والتستوستيرون الحر وبروتين سي التفاعلي، إلى جانب انخفاض تراكيز الهرمون المنشط للجريب .($p<0.001$) أظهر تحليل ملف الدهون مستويات أعلى بشكل كبير من الكوليسترول الكلي والبروتين الدهني منخفض الكثافة، مع تراكيز أقل من البروتين الدهني عالي الكثافة في مريضات متلازمة المبيض متعدد الكيسات .($p<0.05$) حدد التحليل الجيني ثلاثة أنماط جينية لـ ApoE (ε3ε3 ، ε2ε3، (ε3ε4، مع كون ε3ε3 الأكثر انتشاراً في كلا المجموعتين. لم تُلاحظ فروق كبيرة في توزيع الأنماط الجينية أو الأليلات لـ ApoE بين مريضات متلازمة المبيض متعدد الكيسات والضوابط .($p>0.05$)ومع ذلك، داخل مجموعة متلازمة المبيض متعدد الكيسات، أظهر حاملو الأليل ε4 ارتفاعاً كبيراً في الكوليسترول الكلي ($p=0.039$) والدهون الثلاثية ($p=0.013$) ومستويات البروتين الدهني منخفض الكثافة جداً ($p=0.026$) مقارنة بحاملي ε2 و ε3 . **الاستنتاج:** لا يبدو أن تعدد الأشكال الجينية لجين ApoE يؤثر بشكل كبير على القابلية للإصابة بمتلازمة المبيض متعدد الكيسات، حيث كانت توزيعات الأنماط الجينية متشابهة بين المريضات والضوابط. ومع ذلك، قد تؤثر متغيراتApoE ، وخاصة الأليلε4 ، على شدة الخلل الأيضي في النساء المصابات بمتلازمة المبيض متعدد الكيسات المؤكدة، مما قد يؤثر على تقسيم مخاطر القلب والأوعية الدموية ونُهج الإدارة العلاجية.

الكلمات المفتاحية: أبوليبوبروتين إي، تعدد الأشكال الجينية، استقلاب الدهون، متلازمة المبيض متعدد الكيسات.