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Genetic variation in the vitamin D receptor gene and vitamin D Serum levels in Egyptian women with polycystic ovary syndrome

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Obesity, insulin resistance, and hyperandrogenism are considered crucial parameters of polycystic ovary syndrome (PCOS) which might be related to vitamin D metabolism. The aim of this study was to investigate the associations between polymorphisms (TaqI and ApaI) in the vitamin D receptor gene (VDR) and PCOS among Egyptian women. We aimed also to elucidate the impact of these polymorphisms on vitamin D level, hormonal and metabolic parameters of PCOS. One hundred and fifty Egyptian women with PCOS and 150 unrelated controls were enrolled in this study. Polymorphisms of VDR Taq-I T/C (rs731236) and Apa-I A/C (rs7975232) gene were genotyped using polymerase chain reaction restriction fragment length polymorphism (PCR-RFLP). Serum 25hydroxy vitamin D [25(OH) D] levels were measured by high-performance liquid chromatography. PCOS women had significantly lower levels of 25(OH) D compared to healthy women. Our results revealed that Taq-I CC genotype and C allele were associated with increased risk of PCOS, while the Apa-I polymorphism was not. Haplotype Taq-I C/ Apa-I C was associated with a higher PCOS risk more than controls. Moreover, there was a significant decrease of 25(OH) D levels in carriers of haplotype Taq-I C/ Apa-I C (with variant alleles) compared to the noncarriers. Results showed also that there was an obesity-VDR Taq-I genotypes interactions. These results suggested that, VDR Taq-I gene polymorphism is associated with increased risk of PCOS in Egyptian women.

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