World Academy of Science, Engineering and Technology
International Journal of Medical and Health Sciences
Vol:9, No:12, 2015

Genetic Polymorphism in the Vitamin D Receptor Gene and 25-Hydroxyvitamin D Serum Levels in East Indian Women with Polycystic Ovary Syndrome

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Abstract : Background: Polycystic ovary syndrome (PCOS) is the most common metabolic abnormality such as changes in lipid profile, diabetes, hypertension and metabolic syndrome occurring in young women of reproductive age. Low vitamin D levels were found to be associated with the development of obesity and insulin resistance in women with PCOS. Variants on vitamin D receptor (VDR) gene have also been related to metabolic comorbidities in general population. Aim: The aim of this case-control study was to investigate whether the VDR gene polymorphisms are associated with susceptibility to PCOS. Methods: Women with PCOS and a control group, all aged 16-40 years, were enrolled. Genotyping of VDR Fok-I (rs2228570), VDR Apa-I (rs7975232) as well as GC (rs2282679), DHCR7 (rs12785878) SNPs between groups were determined by using direct sequencing. Serum 25-hydroxyvitamin D [25(OH)] levels were measured by ELISA. Results: Mean serum 25(OH)D in the PCOS and control samples were 19.08±7 and 23.27±6.03 (p=0.048) which were significantly lower in PCOS patients compared with controls. CC genotype of the VDR Apa-I SNP was same frequent in PCOS (25.6%) and controls (25.6%) (OR: 0.9995; 95%CI: 0.528 to 1.8921; p= 0.9987). The CC genotype was also significantly associated with both lower E2 (p=0.031) and Androstenedione levels (p=0.062). We observed a significant association of GC polymorphism with 25(OH)D levels. PCOS women carrying the GG genotype (in GC genes) had significantly higher risk for vitamin D deficiency than women carrying the TT genotype. Conclusions: In conclusion, data from this study indicate that vitamin D levels are lower, and vitamin D deficiency more frequent, in PCOS than in controls. The present findings suggest that the Apa-I, Fok-I polymorphism of the VDR gene is associated with PCOS and seems to modulate ovarian steroid secretion. Further studies are needed to better clarify the biological mechanisms by which the polymorphism influences PCOS risk.

Keywords : vitamin D receptor, polymorphism, vitamin D, polycystic ovary syndrome
Conference Title : ICOG 2015 : International Conference on Obstetrics and Gynaecology
Conference Location : Bangkok, Thailand
Conference Dates : December 17-18, 2015