ABSTRACT

Polycystic ovary syndrome is a common endocrine disorder in women of reproductive age. Hyperandrogenism is one the characteristics of polycystic ovary syndrome (PCOS) which may be related to the activity of androgen receptor (AR). Androgen receptor plays a vital role for development of ovarian follicles via binding with androgen. In order to identify the association of androgen level and genetic variation in lean and obese PCOs subjects, firstly, DNA of 100 PCOS and 100 controls was extracted to detect the polymorphism in AR gene. Sequences analysis was performed to confirm the polymorphism in lean and obese Pakistani population. The genotype frequency and allele frequency was calculated in women with PCOS and control for rs12014709. The genotype frequency for ''GG'' in obese PCOS was 46.15%, in Lean PCOS was 39.58% and in control was 27% ($p = 0.001$, OR = 6.57). For TT genotype frequency in PCOS obese and lean was respectively 9.6% and 18.75% while it was 27% in control (($p = 0.001$). For homozygous GT genotype frequency in PCOS obese was 44.23% and in lean was 41.66%, however in control was 26%. The allele frequency for G allele in PCOS was 71% and 50.5% in control. For T allele, frequency for PCOS was 33% while 49.5% in control. The higher level of testosterone was measured in obese patients that were 1.67 ng/ml. Lean patients also showed higher concentration of testosterone which was 1.1 ng/ml. The lower level of testosterone found in controls was 0.32 ng/ml. There was significant difference in androgens concentration among PCOS and controls ($p=0.001$).

Keywords: PCOS, Polymorphism, Testosterone, Androgen receptor