Association of vitamin D receptor gene polymorphisms with end-stage renal disease and the development of high-turnover renal osteodystrophy in a Chinese population

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ABSTRACT. Two single nucleotide polymorphisms (SNPs; TaqI and ApaI) in the vitamin D receptor (VDR) gene have been identified as risk factors for the progression of end-stage renal disease (ESRD). The purpose of our study was to confirm the reported association of these two SNPs with ESRD risk and progression of renal osteodystrophy in a Chinese Han population. A total of 452 ESRD patients and 904 matched-pair controls (based on age, gender, and body mass index) were included. Identification of VDR gene polymorphisms was performed using the polymerase chain reaction-restriction fragment length polymorphism method with TaqI and ApaI restriction enzymes.
There was no association of the TaqI polymorphism with ESRD risk. However, significant associations were seen between ApaI (rs7975232) polymorphism and ESRD risk in the heterozygote model (AC/ AA; P = 0.002; OR = 1.4, 95%CI = 1.14-1.83), homozygote model (CC/AA; P = 0.007; OR = 1.8, 95%CI = 1.17-2.85) genotypes for rs7975232, allelic model (P < 0.001; OR = 1.4, 95%CI = 1.15-1.64), dominant model (P = 0.001; OR = 1.5, 95%CI = 1.19-1.87), and recessive model (P = 0.046; OR = 0.6, 95%CI = 0.42-1.00) between cases and healthy controls. Moreover, we found a significant correlation between the genotype and allele distribution of ApaI and intact parathyroid hormone (iPTH) levels, where allele C carriers have increased iPTH levels. The ApaI polymorphism in the VDR gene appears to be a susceptibility locus for ESRD in Chinese individuals, and allele C carriers may have an increased risk of high-turnover renal osteodystrophy.

Key words: ApaI; TaqI; End-stage renal disease; Polymorphism; Renal osteodystrophy