Relationship between estrogen receptor 1 gene polymorphisms and postmenopausal osteoporosis of the spine in Chinese women


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ABSTRACT. The purpose of this study was to evaluate single nucleotide polymorphism (SNP) variants of the estrogen receptor 1 gene (ESR1) at rs2234693 and rs9340799, as well as to investigate the relationship between ESR gene polymorphisms and postmenopausal osteoporosis (OP) of the spine in Chinese women. We recruited 198 postmenopausal women with OP and 276 healthy women between May 2012 and September 2015 in Zhongshan Hospital. Dual energy x-ray absorptiometry was used to measure the bone mineral density (BMD) of the lumbar vertebrae in all subjects. In addition, PCR-restriction fragment length polymorphism based analysis was conducted to identify the genotypes of ESR1. The distribution of ESR1 in the osteoporosis group and the control group was determined; the relationship between ESR polymorphisms and BMD was analyzed. The distributions of BMD were: TT < TC < CC, GG < AG < AA. The TT, TTGG, and TCGG genotypes were found to be lower as compared to the other genotypes. Stratified analysis suggested that the TT genotype and the combined genotypes TTGG and TCGG were significantly higher in the OP group as compared to the control group (P < 0.01). Therefore, ESR1
polymorphisms at rs2234693 and rs9340799 may be associated with OP, and could be used as markers to screen those with high risks to postmenopausal OP in Chinese women.

**Key words:** Osteoporosis postmenopausal; Bone mineral density; Estrogen receptor 1 polymorphisms; Spine