عنوان الوثيقة: العوامل الوراثية المتنوعة لمجسات الإستروجين ألفا والكثافة الكتلية العظمية.

الموضوع: أمراض الغدد الصماء والعوامل الوراثية.

لغة الوثيقة: الإنجليزية.

المستخلص:

Objectives : To study the frequency of ER-α gene polymorphisms *Xba* I and *Pvu* II in relation to bone mineral density (BMD) and serum estradiol (E_2) variation in Saudi postmenopausal women with and without osteoporosis.

Subjects and Methods : A total of 600 Saudi postmenopausal women (age > 50 years) with osteoporosis (n = 300) and as compared with age-matched women with normal BMD (n = 300) were studied. Women were genotyped by restriction fragment length polymorphism (RFLPs) of ER-α and BMD [at lumbar spine (L_1-L_4) and femoral neck] were determined by dual energy X-ray absorptiometry (DXA).

Results : Women with the genotypes XX and PP exhibited higher BMD values at both the lumbar spine (by 11.6% and 8.2%, $P < 0.05$) and the femoral neck (12.5% and 4.9%, $P < 0.05$), respectively, than those with xx and pp genotypes. Regression analysis showed that women with xx and pp genotypes had a relatively accelerated decrease in BMD values with age at both the lumbar spine ($P < 0.001$) and femoral neck ($P < 0.001$) sites. The XX genotypes were significantly more prevalent ($P < 0.001$) among women with normal BMD (35.7%) and xx genotypes in women with osteoporosis (23.3%) within the group, respectively. The frequency of PP genotype was higher in women with normal BMD (28.7%) whereas pp genotype was higher in women with osteoporosis (46.3%). The mean E_2 was significantly low in women with pp and xx genotypes as compared with that in women with PP and XX genotypes ($P < 0.000$), respectively: this is more significant in women with osteoporosis. The relative risk associated with the presence of a particular genotype was 2.36 ($P < 0.001$) for xx and 1.66 ($P < 0.06$) for pp genotype, respectively.

Conclusions : Genetic variations at ER-α gene locus are associated with BMD values (lumbar spine and femoral neck) in Saudi postmenopausal women which may contribute to the changes of bone loss with age and osteoporosis with significantly low BMD and E_2 values obtained in women with pp and xx genotypes.

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