Polymorphisms in CYP17, COMT, and ESR1 genes in women after menopause and association with bone mineral density

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Received March 16, 2015
Accepted July 23, 2015
Published December 1, 2015
DOI http://dx.doi.org/10.4238/2015.December.1.32

ABSTRACT. In this study, we evaluated genetic factors related to the mineral density during post-menopause. We evaluated 110 women in the first 5 years post-menopause, without previous hormone replacement therapy. Cytochrome P450 17 (CYP17) (rs743572), catechol-O-methyl transferase (COMT) (rs4680), and estrogen receptor 1 (ESR1) (rs9322331) were examined for the presence of polymorphisms. Clinical data were collected by anamnesis; all patients had the osseous densitometry examined using a lunar instrument to determine mineral osseous densitometry in the lumbar column (L2-L4). CYP17, COMT, and ESR1 genotyping was carried out by polymerase chain reaction with DNA collected from buccal swabs. The average age was 51.96 years. The average weights of the patients in control and osteopenia groups were 70.25 ± 12.00 and 62.45 ± 11.64, respectively (P = 0.001) and body mass index (P = 0.006; control: 29.43 ± 5.25; osteopenia: 26.72 ± 4.57). Related to CYP17 polymorphisms, 28.18% of women were TT (wild-type homozygous), 60%
were TC (heterozygous), and 11.82% were CC (mutated homozygous). Related to COMT polymorphisms, 53.64% of women were GG (wild-type homozygous), 37.27% were GA (heterozygous), and 9.09% were AA (mutated homozygous). Related to ESR1, 53.64% of women were CC (wild-type homozygous), 40.91% were CT (heterozygous), and 5.45% were TT (mutated homozygous). The ESR1 variant allele was significantly higher in the osteopenia group when compared with women in the normal group (P = 0.02). ESR1 may be associated with low mineral osseous densitometry, while CYP17 and COMT gene polymorphisms were not associated with mineral osseous densitometry.

**Key words:** Catechol-O-methyl transferase; Cytochrome P450 17; Estrogen receptor 1; Osteoporosis; Polymorphism