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

C.G. Gonçalves[1], B.C. Almeida[1], C.M. Camargo-Kosugi[1,2], A.M.M. Costa[1], I.D.C.G. Silva[1] and M.A. Haidar[1]

[1]Laboratório de Ginecologia Molecular e Proteômica, Departamento de Ginecologia, Universidade Federal de São Paulo, São Paulo, SP, Brasil
[2]Departamento de Otorrinolaringologia e Cirurgia de Cabeça e Pescoço, Universidade Federal de São Paulo, São Paulo, SP, Brasil

Corresponding author: C.M. Camargo-Kosugi
E-mail: cintiakosugi@gmail.com

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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

