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Association analysis of vitamin D receptor gene polymorphisms and bone mineral density in postmenopausal Mexican-Mestizo women

A. González-Mercado[1,2], J.Y. Sánchez-López[1,2], J.A. Regla-Nava[1,a], J.I. Gámez-Nava[3,4], L. González-López[5,6], J. Durán-González[1,2], A. Celis[3,6], F.J. Perea-Díaz[1,2], M. Salazar-Páramo[4,7] and B. Ibarra[1,2]

[1]División de Genética, Centro de Investigación Biomédica de Occidente, IMSS, Guadalajara, Jalisco, México
[2]Doctorado en Genética Humana, Universidad de Guadalajara, Guadalajara, Jalisco, México
[3]Unidad de Investigación Médica en Epidemiología Clínica, Hospital de Especialidades, CMNO, IMSS, Guadalajara, Jalisco, México
[4]Doctorado en Farmacología, Departamento de Fisiología, CUCS, Universidad de Guadalajara, Guadalajara, Jalisco, México
[5]Servicio de Reumatología del HGR110 IMSS, Guadalajara, Jalisco, México
[6]Doctorado en Salud Pública, Departamento de Salud Pública, CUCS, Universidad de Guadalajara, Guadalajara, Jalisco, México
[7]División de Investigación en Salud, Hospital de Especialidades, CMNO, IMSS, Guadalajara, Jalisco, México

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[a]Present address: Department of Molecular and Cell Biology, Centro Nacional de Biotecnología (CNB-CSIC), Madrid, Spain
Corresponding author: B. Ibarra
E-mail: bibarra@mail.udg.mx

ABSTRACT. We investigated associations between vitamin D receptor (*VDR*) gene polymorphisms, *Fok*I T>C (rs2228570), *Bsm*I G>A (rs1544410), *Apa*I G>T (rs7975232), and *Taq*I T>C (rs731236), with bone mineral density (BMD) in postmenopausal Mexican-Mestizo women. Three hundred and twenty postmenopausal women participated,

who were classified according to World Health Organization criteria as non-osteoporotic (Non-OP; N = 88), osteopenic (Opn; N = 144), and osteoporotic (OP; N = 88). BMD measurements at the lumbar (L1-L4) spine and at the left and right femoral neck were obtained by dual-energy X-ray absorptiometry. Single nucleotide polymorphisms (SNPs) were genotyped using real-time polymerase chain reaction and TaqMan probes. Genotype and allelic frequencies of the 4 *VDR* SNPs were similar among the 3 groups. Polymorphic allele frequencies were as follows: *Fok*I (C) 0.53, 0.49, 0.56; *Bsm*I (A) 0.26, 0.22, 0.23; *Apa*I (T) 0.43, 0.39, 0.44; *Taq*I (C) 0.27, 0.22, 0.23 for the Non-OP, Opn, and OP groups, respectively. Although no associations were found between the SNPs and BMD, based on the putative function of the *Fok*I SNP, we constructed, for the first time, the haplotype with the 4 *VDR* SNPs, and found that the CGGT haplotype differed between the Non-OP and OP groups (21.8 *vs* 31.8%, $P < 0.05$). The risk analysis for this haplotype was nearly significant under the dominant model (OR = 1.783, 95%CI = 0.98-3.25, P = 0.058). This result suggests a possible susceptibility effect of the C allele of the *Fok*I SNP for the development of osteoporosis in postmenopausal Mexican-Mestizo women.

Key words: Bone mineral density; Postmenopausal women; Osteoporosis; *VDR* polymorphisms