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Objectives. Osteoporosis is a common disorder with reduced bone mineral density (BMD) and increased susceptibility to fracture. As much as 80% of the normal variation of BMD is influenced by genetic factors. Identifying the specific genes and unfavorable allelic variants underlying peak BMD will help reveal the pathogenesis of osteoporosis and ultimately improve diagnosis, prevention, and treatment strategies of this complex disease. Comparison between ethnic groups permit the identification of risk alleles shared between populations.

Aim. The aim of the study was to evaluate the association between BMD and nine single nucleotide polymorphisms within five osteoporosis predisposition genes in Belarusian and Lithuanian women.

Materials and methods. Case group included women with severe postmenopausal osteoporosis (PMO) (54 Belarusians, average age 58.3±6.2 years, and 28 Lithuanians, aged 74.1±1.2 years), the control group comprised postmenopausal women with the BMD T-score of >–2.5 and without

previous fragility fractures (77 Belarusians, 56.7±7.42 years and 45 Lithuanians, 72.9±0.9 years, $p>0.05$). DNA was extracted from bloodspots dried on special cards (Macherey-Nagel, Germany). Polymorphic sites in osteoporosis susceptibility genes (VDR ApaI, BsmI, TaqI and Cdx2, COL1A1 G2046T, COL1A2 A/G, COL5A1 T/C and T/A and LCT T-13910C gene polymorphisms) were determined using PCR analysis. Significance was assessed using $\chi 2$ test. The differences were considered significant at $p<0.05$.

Results. The analysis of samples from Belarusian women revealed association of VDR ApaI, BsmI and LCT T-13910C gene polymorphisms with PMO. The risk of osteoporosis was 3.3 times higher for the bearers of AA-genotype of VDR ApaI gene polymorphism and 2.6 times higher for B-allele bearers of VDR BsmI, compared to controls ($p<0.05$). The genotyping of Lithuanian women showed that the total frequency of unfavorable risk alleles (predisposing to PMO) in case group (52.1%) was higher comparing to controls (48.6%). No statistically significant difference was found between Lithuanian women with PMO and control group. A statistically significant correlation between VDR ApaI and VDR TaqI risk genotypes and BMD level observed in Belarusian women. The analysis of the relationship between gene polymorphisms and BMD in Lithuanian population revealed a statistically significantly higher BMD levels at lumbar spine in CT heterozygotes of LCT T-13910C gene polymorphism compared to unfavorable CC homozygotes.

Conclusions. In general, the findings of this study reveal the genetic mechanisms, determining decrease of BMD, and gene polymorphisms, which can be considered as markers of predisposition to osteoporosis and used for preventive measures.