DIAGNOSTICS OF GENETIC PREDISPOSITION TO OSTEOPOROSIS IN BELARUSSIAN POSTMENOPAUSAL WOMEN

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Objectives. The evaluation of the molecular and genetic causes of osteoporosis is quite an actual task. Variations of osteoporosis in the population are associated with the

interaction between genotype and environment. Development of diagnostics system of genetic markers testing may enable early identification of risk groups to perform preventive measures.

Aim. The aim of the study was to analyze the association of polymorphisms of Vitamin D receptor (VDR) gene, type I collagen gene (COL1A1) and lactase gene (LCT) with severe postmenopausal osteoporosis and with bone mineral density.

Materials and methods. A total of 54 women with severe postmenopausal osteoporosis (average age 58.3±6.2 years) were included in this study. Control group consisted of 77 Caucasian patients recruited from City Center of Osteoporosis Prevention (Minsk, Belarus) without osteoporosis, corresponding to the main group by sex and age (all women, average age 56.7±7.42 years, the difference is non-significant). The data of medical histories were obtained and anthropometrical measurements (body weight, height) were performed. Dual-energy X-ray absorptiometry (iDXA, GE Lunar) was used to measure bone mineral density (BMD) and bone mineral content (BMC). DNA was extracted from bloodspots dried on special NucleoSafe cards (Macherey-Nagel, Germany). Polymorphic sites in osteoporosis predisposition genes (ApaI, BsmI, TaqI and Cdx2 polymorphisms of VDR gene, G2046T polymorphism of COL1A1 gene and T-13910C polymorphism of LCT gene) were determined using polymerase chain reaction (PCR) analysis using specially designed primers. Statistical analysis was performed using STATISTICA 10 for Windows.

Results. The data shows that VDR ApaI, BsmI and LCT T-13910C polymorphisms are likely to influence the risk of postmenopausal osteoporosis and make the greatest contribution to its development in Belarusian population. For the bearers of AA-genotype of VDR ApaI gene polymorphism, the risk of bone fracture was 3.3 times higher, and for B-allele bearers of VDR BsmI, the risk of osteoporotic fractures was 2.6 times higher if compared to controls. A statistically significant correlation between VDR ApaI and VDR TaqI risk genotypes and

BMD level was observed.

Conclusions. The findings of this study suggest that at least VDR ApaI, BsmI and TaqI and LCT T-13910C polymorphisms are likely to influence the risk of postmenopausal osteoporosis and make the greatest contribution to its development in Belarusian population. At the same time, polymorphisms of the VDR Cdx2 and COL1A1 G2046T demonstrated non-significant association with osteoporosis, and further molecular and genetic analysis is required. Screening of these genetic markers may enable early identification of risk groups to perform preventive measures. Physicians of various specialties, for their successful practice, must have complete knowledge about the influence of the polymorphisms in human genes on the development of pathological processes.

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DIAGNOSTYKA W KIERUNKU PREDYSPOZYCJI GENETYCZNYCH DO OSTEOPOROZY U BIAŁORUSKICH KOBIET W WIEKU POMENOPAUZALNYM

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