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VITAMIN D RECEPTOR (VDR) GENE POLYMORPHISMS AND THEIR ASSOCIATION WITH ASEPTIC NECROSIS OF THE FEMORAL HEAD: A COMPARATIVE CROSS-SECTIONAL ANALYSIS

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Relevance. Aseptic necrosis of the femoral head (ANFH) accounts for 5% of all orthopedic hip pathologies. Around 5 000-20 000 new cases are diagnosed annually in Europe and the United States. The social significance of this problem stems from the potential for disability in young patients due to the lack of early diagnosis and effective treatment. The medical importance of this study arises from the difficulty of early detection of the pathology in the early stages of the disease, the increased cost of patient treatment, and the requirement for hip replacement.

Aim: the study examines the levels of total vitamin D and osteocalcin in the blood of patients with aseptic necrosis of the femoral head and explores the polymorphism of the VDR gene (rs2228570) to identify a genetic predisposition to the development of aseptic necrosis of the femoral head.

Materials and methods. This comparative cross-sectional study was conducted at the Department of Traumatology, Orthopedics, and Military Field Surgery, Grodno City Clinical Emergency Hospital, during the period of June 2024 to March 2025. The patients were divided into two groups: aseptic necrosis of the femoral head (n=44) and hip osteoarthritis (n=30). Inclusion criteria included diagnosis of M87.0 (unilateral or bilateral aseptic necrosis of the femoral head), M16.0 or M16.1 (unilateral or bilateral stage 3 hip osteorthritis by Kellgren-Lawrence) and informed consent. Exclusion criteria encompassed lack of informed consent from patients, other joint diseases, acute infections, malignancies, and severe renal or hepatic failure. Blood samples were collected for biochemical analysis of total vitamin D and osteocalcin levels using ELISA kits, following standardized protocols, along with samples for genetic testing.

Results and their discussion. This study compared serum Vitamin D and osteocalcin levels, and VDR gene Met1Thr (rs2228570) polymorphisms in patients with aseptic necrosis of the femoral head (ANFH, n=41-42) and hip osteoarthritis (coxarthrosis, n=27-30). Both groups exhibited severe vitamin D deficiency (median 3.79 ng/mL in coxarthrosis and 2.82 ng/mL in ANFH) with no significant difference in Vitamin D levels ($p>0.1$), even after excluding outliers. Serum osteocalcin levels were profoundly reduced in both groups (median 1.31 ng/mL for ANFH, 0.85 ng/mL for coxarthrosis), indicating marked suppression of bone formation; no significant intergroup differences were found ($p>0.2$). The genotypic distributions of the VDR Met1Thr polymorphism showed no statical significance among the groups ($\chi^2=2.081$, $p=0.353$). However, the carriers of the G (Thr) allele showed a non-significant pattern toward increased ANFH risk (OR=2.16, 95% CI 0.74–6.31), this was statistically inconclusive. These findings highlight that both ANFH and hip osteoarthritis share severe vitamin D deficiency and impaired bone metabolism, while VDR Met1Thr polymorphism is not significantly correlated with ANFH in this cohort. The results emphasize the significance of monitoring the vitamin D status and bone turnover in these patients, even though genetic risk stratification through VDR genotyping requires further studies.

Conclusions. This study assessed total vitamin D and osteocalcin levels, and the VDR gene (rs2228570, C/T) polymorphism, to identify genetic predisposition to femoral head osteonecrosis.