SECTION 19.
SCIENCES MÉDICALES ET SANTÉ PUBLIQUE

DOI 10.36074/logos-31.10.2025.041

BsmI POLYMORPHISM OF THE VITAMIN D RECEPTOR GENE IN CHILDREN OF THE UKRAINIAN POPULATION WITH TYPE 1 DIABETES

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Vitamin D (vit. D) and the vitamin D receptor (VDR) gene affect human health, as they regulate the expression of genes that control calcium homeostasis, energy metabolism, and immune processes [1]. The involvement of vitamin D in the etiopathogenesis of type 1 diabetes (T1DM) cannot be ruled out [2,4]. The results of studies conducted in some populations suggest that the BsmI polymorphism may be a risk factor for the development of diabetes, but this requires further research [3,5,6]. In Ukraine, no studies have been conducted on the association between the BsmI polymorphism of the VDR gene and the content of vit. D in children with T1DM.

The objective of this article was to study the BsmI (rs1544410) polymorphism of the vit. D receptor gene in children of the Ukrainian population and assess its association with T1DM. **Materials and methods.** 50 children and adolescents (19 boys and 31 girls) with T1DM aged 1,75 to 17,08 years (mean age 12,1±3,7 years) were

examined. Glycated hemoglobin, serum 25(OH)D were determined by immunochemiluminescence method, and molecular polymorphism BsmI (rs1544410) of the *VDR* gene in children with T1DM by polymerase chain reaction method.

Results. The examined children were divided into three groups: Group I – 26 children (52%) – with the GA genotype of the BsmI polymorphism (rs1544410) of the *VDR* gene; Group II – 15 children (30%) – with the GG genotype and Group III – 9 children (18%) – with the AA genotype. In children with T1DM and the pathological homozygous (AA) polymorphism, glycemic control was classified as high-risk for life, whereas children in the other groups demonstrated suboptimal glycemic control. Vit. D deficiency was detected in patients with the heterozygous (GA) and pathological homozygous (AA) genotypes (42,1±16,8 nmol/L and 40,2±11,3 nmol/L, respectively). In children with the normal (GG) genotype of the BsmI (rs1544410) *VDR* gene polymorphism, vit. D insufficiency was observed (54,4±12,6 nmol/L). The GA genotype of BsmI (rs1544410) of the *VDR* gene (52%) was dominant in the examined children, which was associated with vit. D deficiency. The GG genotype (66%) was dominant in the control group. In the control group, the GG genotype predominated (66%). Children with the GA genotype had a significantly increased risk of developing T1DM – OR = 3,55 (95% CI 1,48–8,50; $p < 0,0005$), whereas the GG genotype was protective against T1DM – OR = 0,22 (95% CI 0,09–0,52; $p < 0,005$).

Conclusions. The results of this study demonstrate that most patients with T1DM had hypovitaminosis D - vit. D deficiency was detected in 34 (68,0%) children, vit. D insufficiency in 15 (30,0%) children, and optimal vit. D supply in only one (2,0%) patient. The GA genotype of the BsmI (rs1544410) *VDR* polymorphism predominates in children with T1DM. In pathological homozygotes of the BsmI (rs1544410) *VDR* polymorphism, unsatisfactory glycemic control was observed against the background of vit. D deficiency. A significant risk of developing T1DM was confirmed in children with the heterozygous polymorphism. It is advisable to determine the BsmI (rs1544410) polymorphism of the *VDR* gene in children with T1DM associated with vit. D deficiency and monitor vit. D supplementation.

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