

Abstract

In this work we chose to study genes whose protein products significantly affect calcium-phosphate metabolism in kidneys and are hypothesized to be involved in the pathogenesis of diabetes as well as kidney damage.

Vitamin D receptor (*VDR*) belongs to a large family of nuclear receptors and transcription factors. *VDR* is expressed in many cells in different tissues and activation of this receptor by its ligand 1,25 - dihydroxyvitamin D₃ affects expression of many genes. Using restriction endonucleases *Bsm I*, *Fok I*, *Taq I* and *Apa I*, there were identified polymorphic sites in *VDR* gene that were found to be associated with certain chronic diseases, like type 1 and type 2 diabetes and lung carcinoma.

Parathyroid hormone is one of the most important regulators of calcium and phosphate metabolism. Allelic polymorphism of *PTH* gene involves B/b and D/d alleles that can participate in pathogenesis of kidney complications as well.

Aims: We aimed to determine whether there is a difference in frequency of certain combinations of *VDR* and *PTH* genotypes between group of diabetic patients with kidney complications and diabetic patients without kidney complications. We also assessed influence of single-nucleotide polymorphisms at *VDR* gene mRNA expression.

Methods: DNA was isolated by salt-extraction method and specific fragments of *VDR* and *PTH* genes were amplified using PCR. Amplified products were digested by allele-specific restrictases *Bsm I*, *Apa I*, *Taq I*, *Fok I* (*VDR* gene) and *Bst BI* and *Dra II* (*PTH*). The genotype of individual was determined on the basis of length of restriction fragments. Assessment of *VDR* RNA levels in full blood was performed using Real-Time PCR.

Results: The BBDD genotype seems to be predisposing factor for development of type 1 diabetes, but not for development of diabetic nephropathy. The bbDd genotype seems to have protective role in development of diabetes and its complications.