



Vitamin D Receptor Gene
Polymorphisms and Breast Cancer
Risk in Kazakhstan

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Abstract

Introduction: The steroid hormone 1,25-dihydroxyvitamin D₃ is thought to protect against breast cancer. The activity of 1,25-dihydroxyvitamin D₃ is mediated via the vitamin D receptor (VDR), and a number of polymorphisms in the *VDR* gene have been identified. These result in distinct genotypes, some of which may alter susceptibility to breast cancer. Two common single nucleotide polymorphisms (SNP) in the *VDR* gene (*VDR*), rs1544410 (BsmI) and rs2228570 (FokI), have been inconsistently associated with breast cancer risk. Increased risk has been reported for the FokI ff genotype, which encodes a less transcriptionally active isoform of *VDR*. A reduced risk has been reported for the BsmI BB genotype which may influence *VDR* mRNA stability.

Aim: We have investigated whether specific *VDR* gene polymorphisms are associated with breast cancer risk in Kazakhstan women.

Material and Methods: In a case–control study, female breast cancer patients (315) and a female control group ($n=604$) were tested for two *VDR* polymorphisms. Statistical analysis was conducted using SPSS19.0.

Results: The *VDR* rs2228570 (FokI) polymorphism was associated with an increased occurrence of BC [rs2228570 (folk) ff vs. FF genotype: OR=1.71; 95% CI=1.21-2.43]. No association was noted between rs1544410 (BsmI) BB and breast cancer risk [OR=0.68; 95% CI=0.49-0.95].

Conclusion: Although the factors that increase breast cancer susceptibility remain uncertain, future large studies should integrate genetic variation in *VDR* with biomarkers of vitamin D status. Additional testing on the effect of varying genotypes on the functional mechanisms of the *VDR* could help to improve future testing and treatment of woman at risk for breast cancer.

Keywords: *breast cancer, gene polymorphysm, vitamin D receptors, genotype suseptibility*

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