



FREQUENCY OF BSMI POLYMORPHISM OF VITAMIN D RECEPTOR GENE AND ITS ASSOCIATION WITH 25-HYDROXYVITAMIN D LEVELS

Huseyin Babur¹, Ahmet Kaya², Elif Turan², Ilker Polat³, Mustafa Kulaksızoğlu², Mahmut Selman Yıldırım⁴, Aysegül Zamani⁴

1-Dr Vefa Tanır Iğın State Hospital Department of Internal Medicine, 2- Necmettin Erbakan University Meram Faculty of Medicine Department of Endocrinology and Metabolism, 3-Afyonkarahisar State Hospital Department of Internal Medicine, 4- Necmettin Erbakan University Meram Faculty of Medicine Department of Medical Genetics

INTRODUCTION

Vitamin D deficiency has become a major public health problem. It's known that the effects of vitamin D are made by its receptors. Vitamin D receptor (VDR) is a member of nuclear receptors type 1 such as androgen, estrogen, progesterone and glucocorticoid receptor. In several studies carried out up to 30 different cells and tissues of the vitamin D receptor (VDR) have shown the presence. In many studies ongoing malignancies and their relationship with Vitamin D and VDR are being investigated. VDR gene polymorphisms in susceptibility to different diseases demonstrated in many studies. The polymorphisms of VDR are ApaI, TaqI, FokI and BsmI. BsmI is the most extensively studied polymorphism. In a study conducted for osteoporosis BB genotype was reported to be low risk indicator [1].

In our study, the aim is to show the frequency of VDR BsmI gene polymorphisms and to show the relationship between polymorphisms and vitamin D levels.

METHOD

All population were divided into two groups which are 100 osteoporotic and 100 non-osteoporotic. The demographic characteristics were recorded. In blood samples vitamin D, calcium, phosphorus, alkaline phosphatase; in urine samples urinary calcium, urinary creatinine were studied. In addition, approximately 2 cc of peripheral blood sample was taken for DNA isolation and VDR BsmI polymorphisms were studied by real-time PCR method and comparisons were made between the groups.

RESULTS

In all of population BB genotype 17 %, Bb genotype 50.5 % and bb genotype 32.5 % were found. In osteoporotic grup BB genotype 16 %, Bb genotype 48 % and bb genotype 36 % were found. In non-osteoporotic grup BB genotype 18 %, Bb genotype 53 % and bb genotype 29 % were found. There was no statistically significant difference between groups ($p=0.571$). The level of vitamin D was 18.10 ng/mL in BB genotype, 17.08 ng/mL in Bb genotype and 16.34 ng/mL in bb genotype. There was no statistically significant difference between genotypes ($p=0.717$).

Fig 1: Vitamin D levels in genotypes

	BB	Bb	bb	p
Vitamin D (ng / mL)	18.10 ± 10.77	17.08 ± 10.25	16.34 ± 9.59	0.717

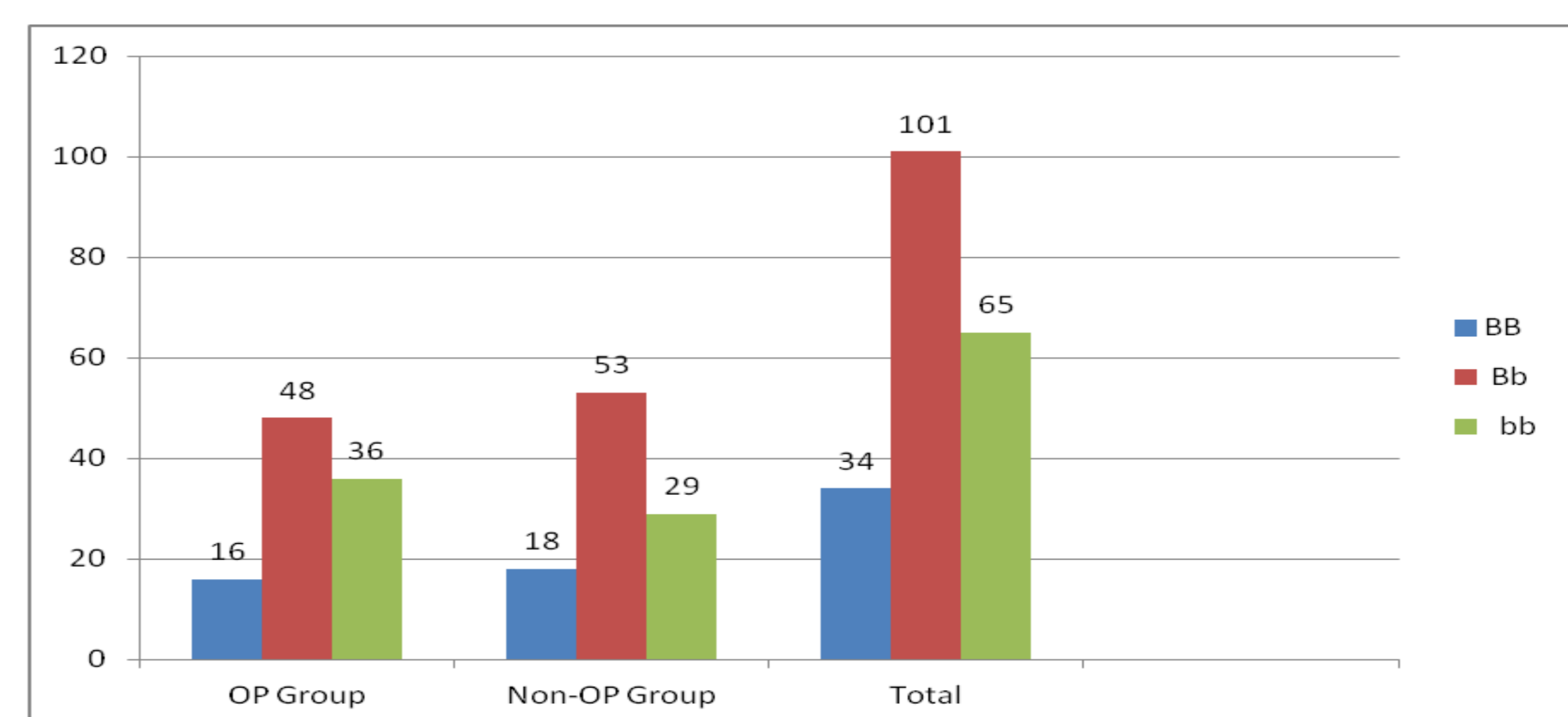


Fig 2: Allels in Groups

DISCUSSION

Has been seen in studies of vitamin D deficiency, especially in developed countries has become a common public health problem [2]. Adequate levels of vitamin D that is currently under discussion but the critical value for the prevention of pathological conditions today minimum qualification as 30 ng / mL was determined [3]. In the absence of susceptibility to some diseases, there was an increase in morbidity and mortality [4]. VDR and vitamin D levels in studies investigating the relationship made and different results were obtained. In a study conducted in Tunisia in asthmatic children has not been shown that the relationship [5], but In a study on children in Japan there was a relationship between VDR and vitamin D deficiency [6]. Examined the relationship between BMD and VDR polymorphisms in the world and in our country, despite the fact that numerous studies conducted have found conflicting results with each other. In a study conducted in Slovenia with femoral neck BMD was found a negative relationship between BsmI polymorphism [7]. On the other hand in a study conducted in the Czech Republic in postmenopausal women with BMD in the relationship between BSM has not been shown [8].

CONCLUSION

In our study there was no statistically significant relationship between genotypes and vitamin D levels and also was no statistically significant relationship of VDR gene BsmI polymorphisms between two groups. Serious vitamin D deficiency was detected in both groups and the whole population.

REFERENCES:

- McKay, J.D., et al., *Vitamin D receptor polymorphisms and breast cancer risk: results from the National Cancer Institute Breast and Prostate Cancer Cohort Consortium*. *Cancer Epidemiol Biomarkers Prev*, 2009. **18**(1): p. 297-305.
- Holick, M.F., *Vitamin D deficiency in 2010: health benefits of vitamin D and sunlight: a D-bate*. *Nat Rev Endocrinol*, 2011. **7**(2): p. 73-5.
- Holick, M.F. and T.C. Chen, *Vitamin D deficiency: a worldwide problem with health consequences*. *Am J Clin Nutr*, 2008. **87**(4): p. 1080S-6S.
- Melamed, M.L., et al., *Serum 25-hydroxyvitamin D levels and the prevalence of peripheral arterial disease: results from NHANES 2001 to 2004*. *Arterioscler Thromb Vasc Biol*, 2008. **28**(6): p. 1179-85.
- Maalmi, H., et al., *The impact of vitamin D deficiency on immune T cells in asthmatic children: a case-control study*. *J Asthma Allergy*, 2012. **5**: p. 11-9.
- Kitanaka, S., et al., *Association of vitamin D-related gene polymorphisms with manifestation of vitamin D deficiency in children*. *Endocr J*, 2012. **59**(11): p. 1007-14.
- Mencej-Bedrac, S., et al., *The combinations of polymorphisms in vitamin D receptor, osteoprotegerin and tumour necrosis factor superfamily member 11 genes are associated with bone mineral density*. *J Mol Endocrinol*, 2009. **42**(3): p. 239-47.
- Zajickova, K., I. Zofkova, and M. Hill, *Vitamin D receptor polymorphisms, bone ultrasound and mineral density in postmenopausal women*. *Aging Clin Exp Res*, 2005. **17**(2): p. 121-4.