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Vitamin D Receptor gene Polymorphism: Association with susceptibility to early-onset breast cancer in Iranian, BRCA1/2-mutation carrier and non-carrier patients

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Mounting evidences support that vitamin D insufficiency or deficiency is a risk factor of breast cancer. Vitamin D receptor (VDR) is expressed in more than 36 cell types in different organs as in cancerous cells. Numerous allelic variants of VDR gene have been identified in human populations. Association of FokI (rs2228570) and BsmI (rs1544410) single nucleotide polymorphisms (SNPs) in VDR gene with the risk of breast cancer have been investigated in several studies, however, the published data are still inconsistent. Here, we investigated BsmI and FokI polymorphisms in Iranian young (≤ 35 years old) breast cancer patient with known BRCA1/2 germline mutations. VDR gene polymorphisms were detected by restriction fragment length polymorphism (RFLP) analysis in a cohort of 203 breast cancer patients and 214 controls from Iran. There was a significant association between the bb and Bb genotypes of the BsmI and the increased risk of breast cancer (OR 1.74, CI 1.06–2.87 and OR 2.08, CI 1.31–3.29, respectively). This association was maintained in the subgroup of BRCA1/2 mutation non carriers (OR 1.90, CI 1.15–3.20 and OR 1.75, CI 1.07–2.87 for bb and Bb genotypes respectively) and in the subgroup of BRCA1/2 mutation non-carriers with a family history of breast and/or ovarian cancer (OR 1.81, CI 1.08–3.05 and OR 1.65, CI 1.00–2.70 for bb and Bb genotypes respectively). None of the FokI homozygous or heterozygous genotypes were associated with the risk of breast cancer. In summary, the BsmI polymorphism of VDR gene may be associated with the risk of breast cancer in Iranian women.

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