Vitamin D Receptor gene polymorphism

The purpose of a study was to investigate the association of Vitamin D Receptor (VDR) gene polymorphisms and VDR levels with lumbar disc degeneration (LDD). TaqMan SNP Genotyping Assay was utilized to probe VDR gene polymorphisms including the FokI (rs2228570), ApaI (rs7975232) and TaqI (rs731236) in 454 patients with LDD and 485 controls. Enzyme-Linked Immunosorbent Assay (ELISA) was used to detect plasma VDR levels. The patients with LDD were divided into three subgroups (subgroup 1: lumbar disc herniation; subgroup 2: lumbar spinal stenosis; subgroup 3: lumbar spondylolisthesis) to further probe the association of plasma VDR levels and VDR gene polymorphisms and LDD. Moreover, immunohistochemistry (IHC) was implemented to evaluate VDR expression in lumbar degenerated disc and normal disc. Allele and genotype frequency of TaqI (rs731236) were significantly different in patients with LDD and controls (all $P < 0.05$). For TaqI polymorphism, the frequencies of T allele were significantly higher in the LDD patients compared with controls (OR = 1.319; 95%CI 1.091 to 1.595; P = 0.004, adjusted (OR = 1.319; 95%CI 1.091 to 1.595; P = 0.004, adjusted OR = 1.383; 95%CI 1.135 to 1.684; P = 0.016). Furthermore, the allele distribution showed a higher frequency of the T allele in the patients with lumbar disc herniation in subgroup 1 (OR = 1.384; 95% CI 1.105 to 1.732; P = 0.004, adjusted OR = 1.319; 95%CI 1.091 to 1.595; P = 0.016). Plasma VDR levels and VDR expression were significantly lower in patients with LDD compared with controls (all $P < 0.05$). Moreover, the TT genotype of TaqI polymorphism was significantly associated with lower plasma VDR levels in patients with LDD (P = 0.002). TaqI (rs731236) polymorphism was associated with a predisposition to LDD. Plasma VDR and VDR expression levels may be the marker for the occurrence and development of LDD [1].

1)

Yang Q, Liu Y, Guan Y, Zhan X, Xiao Z, Jiang H, Wei Q. Vitamin D Receptor gene polymorphisms and plasma levels are associated with lumbar disc degeneration. Sci Rep. 2019 May 24;9(1):7829. doi: 10.1038/s41598-019-44373-2. PubMed PMID: 31127184.