HSR London International Conference on Multidisciplinary Academic Research
(HSR-ICMAR) 2026
https://houghtonstreetreview.com/index.php/conference

DISTRIBUTION PATTERNS OF VDR GENE POLYMORPHISM GENOTYPES IN CHRONIC OBSTRUCTIVE PULMONARY DISEASE

Abdigaffar G. Gadaev
Muborak S. Salaeva
Shoira G. Matrzaeva
Tashkent State Medical University, Uzbekistan
https://doi.org/10.5281/zenodo.22127964

Background. Chronic obstructive pulmonary disease (COPD) is a multifactorial disorder in which genetic factors, along with environmental exposures, may contribute to disease development and clinical course. The vitamin D receptor gene (VDR) is involved in the regulation of immune-inflammatory processes, cell proliferation, and tissue homeostasis. Therefore, different polymorphic variants of the VDR gene are of interest in characterizing the genetic profile of patients with COPD.

Objective. To evaluate the frequency and distribution patterns of genotypes of the VDR gene polymorphisms rs731236 (TaqI), rs7975232 (ApaI), rs1544410 (BsmI), and rs2228570 (FokI) in patients with COPD.

Materials and Methods. Genotypes of four polymorphic loci of the VDR gene — rs731236 (TaqI), rs7975232 (ApaI), rs1544410 (BsmI), and rs2228570 (FokI) — were analyzed in patients with COPD. VDR gene polymorphisms were determined by real-time polymerase chain reaction (Real-Time PCR) using a Rotor-Gene 6000 amplifier (Corbett Research, Australia). The frequencies of homozygous and heterozygous genotypes for each polymorphism were assessed as absolute numbers and percentages.

Results. For the rs731236 (TaqI) polymorphism, the heterozygous TC genotype was the most common and was identified in 53.2% of patients. The TT genotype was observed in 38.1% and the CC genotype in 6.3%; the genotype could not be determined in 2.4% of patients. Thus, the TC genotype represented the predominant variant at this locus.

For the rs7975232 (ApaI) polymorphism, the heterozygous AC genotype was also predominant, occurring in 56.3% of patients. The CC genotype was detected in 23.0% and the AA genotype in 19.8% of patients. The genotype was not determined in 0.8% of cases.

For the rs1544410 (BsmI) polymorphism, the frequencies of the GA and GG genotypes were similar, accounting for 46.0% and 43.7%, respectively. The AA genotype was less frequent and was detected in 9.5% of patients, while the genotype was not determined in 0.8%. These findings indicate that the GA and GG variants constituted the predominant genotypes at the BsmI locus among patients with COPD.

The most distinct distribution pattern was observed for the rs2228570 (FokI) polymorphism. The GG genotype predominated, occurring in 62.7% of patients, whereas the GA genotype was identified in 28.6% and the AA genotype in 7.9%. The genotype was not determined in 0.8% of patients.

Overall, the genotype distribution of VDR gene polymorphisms was heterogeneous among patients with COPD. Heterozygous TC and AC genotypes predominated at the TaqI and ApaI loci, respectively; GA and GG genotypes were most frequent at the BsmI locus; and the GG genotype showed a clear predominance at the FokI locus.

Conclusion. Patients with COPD demonstrated a distinct distribution pattern of VDR gene polymorphism genotypes. Heterozygous genotypes predominated for TaqI and ApaI, GA and GG

variants were most frequent for BsmI, and the GG genotype predominated for FokI. These findings may provide a basis for further characterization of the molecular genetic features of COPD. However, comparison with a control group is required before these genotypes can be interpreted as risk or protective factors for COPD.

Keywords: chronic obstructive pulmonary disease, VDR gene, genetic polymorphism, TaqI, ApaI, BsmI, FokI, genotype.