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SINGLE NUCLEOTIDE POLYMORPHISM -936C/T OF THE VEGF GENE AND DIABETIC RETINOPATHY: AN ASSOCIATION STUDY

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Diabetes is one of the most serious challenges to health care worldwide. It is estimated that diabetes affects around 200 million people, and the number of people affected will continue to increase over next years.
Diabetic retinopathy (DR) is a major complication of diabetes and is a leading cause of vision loss and blindness in the western countries. DR is characterized by a gradually progressive alteration in the retinal microvasculature, including blood-retinal barrier breakdown and neovascularization. Hiperglycemia is known to be the primary pathogenic factor in the development of DR and is responsible to activate abnormal biochemical pathways that in turn influence several vasoactive factors, such as the upregulation of vascular endothelial growth factor (VEGF). VEGF plays a key role in DR, acting as a vascular permeabilizing factor, involved in the blood-retinal barrier breakdown, in the earliest stages of the disease, and as an angiogenic factor promoting the neovascularization, at the later stages.
Family, twin and adoption studies have shown that genetic factors are involved in DR. Regarding the importance of VEGF in the pathogenesis of DR, it can be considered an attractive candidate susceptibility gene in DR. Thus,in this study, we investigated the possible implication of the -936C/T polymorphism of the VEGF gene in the pathogenesis of DR.
The population studied consists of 137 Portuguese patients with type II diabetes, 63 patients with DR, and 74 patients without DR. Genomic DNA was extracted from peripheral blood using an enzymatic procedure. The -936C/T polymorphism of the VEGF gene was amplified with a polymerase chain reaction (PCR), and the PCR product was digested with the restriction enzyme NlaIII. In this association study, we did not found statistically significant genetic association between -936C/T polymorphism of the VEGF gene and DR ($\chi 2$=2.284; df=2; p=0.319). Therefore, this study does not support the hypothesis that the -936 C/T polymorphism of the VEGF gene contribute with a minor gene effect to the expression of DR.