MTHFR (C677T AND A1298C) POLYMORPHISMS AND DIABETIC RETINOPATHY

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Introduction: Diabetic retinopathy is a diabetes related micro-vascular complication of the retina. The role of genetics is well explained by the fact that some diabetic individuals do not develop retinal complications even after a long duration of uncontrolled diabetes.

Aim: The present research endeavoured to study the association between diabetic retinopathy and the C677T and A1289C polymorphisms of the MTHFR gene. Thirty-four patients (24 women and 10 men) as well as 50 healthy subjects were recruited for this case-controlled study. Genotyping was accomplished via the PCR-RFLP technique. This is the first study to ascertain the association between these polymorphisms and DR carried out in Tunisian patients.

Results: Our findings suggest the absence of any association between diabetic retinopathy and different genotypes for the MTHFR C677T gene. Nevertheless, a protective effect of the C allele against this disease was demonstrated (p<0.05); whereas, the T allele (p=0.03) conferred a predisposition to DR. Moreover, statistical analysis revealed that the heterozygous (AC) and wild-type (AA) genotypes were statistically significant and accorded a predisposition for (p=0.01) and protection against (p=0.003) disease, respectively. Also, a protective effect of the A allele against this disease was demonstrated (p<10⁻³) while the C allele (p<10⁻³) conferred a predisposition to the pathology.

Conclusion: This study was limited by the small size of subjects; however, there is evidence of an association between the MTHFR 677 and 1298 polymorphisms and diabetic retinopathy. A study involving a larger cohort is required to understand more definitively the role of these mutations in this pathology.