STUDY OF CYP11B2 GENE POLYMORPHISM ASSOCIATION WITH DIABETES AND HYPERTENSION IN PAKISTANI PUNJABI SUBJECTS

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Diabetic Nephropathy (DN) is a severe complication of diabetes and a major cause of morbidity and mortality worldwide, including Pakistan. Several molecular pathways have been linked to DN at the genetic and biochemical levels including RAAS pathway. Hence, a SNP (T/C) in the aldosterone synthase gene (CYP11B2), which is member of RAAS pathway, was studied in Pakistani Punjabi subjects for its association with DN and related parameters. In this study, a total of 206 subjects from Faisalabad, Pakistan were included. Four study groups were: G1: Controls (without diabetes and nephropathy, n= 52), G2: Type 2 Diabetics (without nephropathy, n= 51), G3: Type 2 Diabetics (with Nephropathy, n= 60), G4: Non-diabetics (controls but with nephropathy, n= 43). Blood samples were collected from all the subjects and were analyzed for biochemical and molecular characterization. Taqman genotyping was done for CYP11B2 gene polymorphism (T/C) to identify the genotypes (TT: homozygous dominant; TC: heterozygous; CC: homozygous recessive), and calculate and compare their genotype frequencies among four groups as well with other biochemical and disease phenotypes like nephropathy, hypertension and family history. The genotyping results showed significant difference in the allele frequency between the control (G1) and nephropathy (G4) groups. Moreover, this polymorphism has also an association with diabetes in this study. This is the first report for this SNP in Pakistani diabetes and DN patients.