



Melanocortin 2 Receptor Mutations and Clinical Significance in Case of Cushing Syndrome and Subclinical Cushing Syndrome and Primary Aldosteronism

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ABSTRACT The researchers investigated whether single nucleotide polymorphisms (SNPs) of *M2CR* were associated with the development of adrenocortical diseases including cushing syndrome (CS), subclinical cushing syndrome (SCS) and primary aldosteronism (PA) in Turkish population. Two promoter SNPs [rs1893219 (853A/G) and rs1893220 (759 G/T)] were genotyped in 43 patients with adrenal adenomas. All patients were examined hormonally with dynamic tests. While 22 of the patients had non-functional (NF) adrenal adenomas; 21 of the patients (CS=9, SCS=10, PA=2) had functional adrenal adenomas. In rs1893219 the frequencies of the CC (AA) were found to be 2, 3 and 1 patients in CS, SCS and PA, respectively. In rs1893220 the frequencies of the AA (GG) genotype were found to be 2, 1 and 1 patients in CS, SCS and PA. Also, the C allele frequency of rs1893219 was found increased in the patients with functional adenomas. The results have shown that the *M2CR* gene may contribute to the development of adrenocortical diseases.