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The leptin receptor gene Q223R single nucleotide polymorphism is not associated with overweight / obesity in urban Sri Lankan women

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Genetic composition can predispose individuals to weight-gain in the obesogenic environment prevailing today. Several studies have indicated that genetic variations in the leptin receptor (LepR) gene may contribute to variability in body weight.

In the present study 100 apparently healthy young women (18 - 31 years), recruited from randomly selected Medical Officer of Health (MOH) divisions of the Colombo Municipality were genotyped for the Q223R polymorphism (rs1137100) of the leptin receptor (LepR) gene. Forty four women were overweight /obese (Body Mass Index (BMI): > 25.0 kg/m²) and 56 were normal weight (BMI: 18.4 - 24.9 kg/m²). Buccal cavity swabs were obtained for DNA extraction.

A PCR-RFLP assay with an internal restriction digestion control was developed for genotyping the subjects. Forward (5'-CAGATACCCCTTTAACTGGGTGTCC-3') and reverse (5'-GCTCTCTGAGGTGGGAACT ATGTCTAA-3') PCR primers were designed to amplify a 763 base-pair fragment spanning the Q223R site of LepR gene. The A to G substitution resulting in the R allele, introduces a *MspI* restriction enzyme site. In this assay, cleavage with *MspI* produced two fragments (467 and 296 bp) for the R allele, in the absence of the internal control. When double digestion with *HinfI* internal control is carried out, the R allele produces three fragments (467, 155 and 141 bp) and the Q allele produces two fragments (622 and 141 bp). In both, presence of the 467 bp band was indicative of R allele.

The frequency of R allele in the study sample was 0.41. In the overweight / obese group the R allele frequency was 0.42 and in the normal weight group it was 0.40. The frequency of RR genotype in the subjects was low (0.05). Genotype distribution between the overweight and normal weight subjects were compared by χ^2 test using a R-dominant genotypic model. No significant difference was observed between the two groups.

In conclusion, a PCR-RFLP assay was developed and optimized for identification of the leptin receptor gene Q223R polymorphism. Our results indicate that the LepR Q223R polymorphism is not associated with overweight / obesity in urban Sri Lankan women.