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Functional and structural impact of ATP-binding cassette transporter A1 R219K and I883M gene polymorphisms in obese children and adolescents

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ATP-binding cassette transporter A1 (ABCA1) mediates transport of intracellular cholesterol across the cell membranes. We aimed to investigate the possible association of *ABCA1* gene polymorphisms; R219K (rs2230806; G/A) and I883M (rs2066714; A/G) with overweight/obesity, and to detect whether these SNPs are associated with lipid profile changes in obese children and adolescents in an Egyptian population. The presence of R219K and I883M genetic variants was determined by polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) analysis in 128 unrelated obese children/adolescents and 128 age- and sex-matched controls. Our findings suggested that the heterozygote GA genotype of R219K polymorphism increased the risk of obesity under heterozygous model (OR = 2.84; 95% CI: (1.23-6.55), P =0.014) and dominant model (OR = 2.32; 95% CI: (1.04-5.14), *P* =0.035) compared to control group. This susceptibility could be gender-specific, with higher risk among females. In addition, the A variant was associated with higher degrees of obesity (class II and III) ($P<0.001$). On the other hand, the G variant of I883M polymorphism significantly decreased the risk of obesity under all genetic models ($p<0.05$), with no observed association with body mass index or categories of obesity. However, both SNPs showed significant differences in lipid levels among different genotypes. In conclusion, the study results suggested that R219K and I883M SNPs of *ABCA1* gene may play a role in susceptibility to obesity in our Egyptian population; the former increases the susceptibility and phenotype severity and the latter is protective. Larger epidemiological studies are needed for results validation.

Biography

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