Genetic polymorphisms affecting the phenotypic expression of patients with molecularly defined familial hypercholesterolaemia

The phenotypic expression of patients with heterozygous familial hypercholesterolaemia (FH) is highly variable [1]. The classes of the LDL receptor (LDLR) mutation and other gene polymorphisms as well as environmental factors have been found to affect the lipid phenotype in patients with heterozygous FH. We read with great interest the recently published data by Bertolini et al. that reported several common variants that influence the lipid profile in FH heterozygotes [2]. Interestingly, the authors mentioned that apolipoprotein (apo) E gene polymorphism affected serum LDL-cholesterol and triglyceride but not HDL-cholesterol levels in patients with molecularly defined FH [2]. We have recently reported that apo E gene polymorphism affected HDL-cholesterol levels in 84 unrelated patients with molecularly defined FH [3]. In fact, the presence of the E4 allele was associated with lower HDL-cholesterol levels in patients without carrying this allele [3]. The above association between HDL-cholesterol and apo E polymorphism still reached statistical significance when our study population expanded to 123 unrelated molecularly defined Greek FH patients. We conclude that the effect of apo E gene polymorphism on lipid parameters in patients with heterozygous FH is still controversial and may be related to the genetic and environmental background of each population studied.

References


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