

Abstract

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Apolipoprotein E and methylenetetrahydrofolate reductase genetic polymorphisms in relation to other risk factors for cardiovascular disease in UK Caucasians and Black South Africans.

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OBJECTIVE: Genetic polymorphisms for apolipoprotein E (apo E) and methylenetetrahydrofolate reductase (MTHFR) are believed to modulate risk of coronary heart disease (CHD) acting through regulation of lipid and homocysteine metabolism, respectively. The distributions of apo E and MTHFR alleles in Black South Africans, a population with a low CHD incidence, and UK Caucasians from the Cambridge area, with a higher CHD incidence, were therefore compared.

METHODS: Clinically healthy volunteers (207), including 107 UK Caucasians from the Cambridge area and 100 Black South Africans, participated in the study. Apo E and MTHFR genotypes were determined in all of them. Analyses for serum total cholesterol, LDL cholesterol, HDL cholesterol, triglycerides and plasma fibrinogen were carried out in 65 UK Caucasians and 60 Black South Africans.

RESULTS: The apo E epsilon4 allele, which is associated with elevated CHD risk, was present in 48% of Black South Africans compared to 20.8% of Caucasians ($P < 0.0001$); however, both total and LDL cholesterol levels in Black South Africans were 18-32% lower than in Caucasians with similar apo E genotypes. Hyperhomocysteinemia-causing MTHFR 677T variant was detected in only 20% of Black South Africans (no homozygotes) versus 56% of Caucasians with 12% homozygotes ($P < 0.0001$).

CONCLUSIONS: Our findings suggest that the potentially unfavourable pattern of apo E allele distribution in Black South Africans does not result in increased CHD incidence due to protection by dietary and/or other life style related factors. The exceptionally low frequency of MTHFR mutant homozygotes in this population suggests that this polymorphism should not be regarded as an important CHD risk factor among Black South Africans.

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