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PvuII polymorphic site upstream to the human ApoCIII gene

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SOURCE/DESCRIPTION: A 2.2 kb KpnI-SacI genomic DNA fragment located between the human apoCIII and apoAIV genes (1) was subcloned in pUC19. The KpnI and SacI sites of this fragment are located 2.4 kb and 4.6 kb, respectively, 5' to the transcription start site of the apoCIII gene (2).
POLYMORPHISM: Genomic restriction mapping analysis of human peripheral lymphocyte DNA, using the KpnI-SacI probe, indicated that presence or absence of a PvuII site located 4.4 kb 5' to the start site of the apoCIII gene results in a two allele polymorphism with allelic fragments of 3.6 kb or 4.3 kb respectively.
FREQUENCY: The frequency of this polymorphism among 66 unrelated Mediterraneans and 36 unrelated Blacks was:

Allele	Mediterraneans	Blacks
3.6 kb	0.069	0
4.3 kb	0.931	1

NOT POLYMORPHIC FOR: BamHI, EcoRI, HindIII, BglII and KpnI.
CHROMOSOMAL LOCALISATION: The ApoAI-AIV cluster has been localised to the long arm of human chromosome 11 (1,3).
MENDELIAN INHERITANCE: Co-dominant segregation demonstrated in 30 Nuclear families.
PROBE AVAILABILITY: Probe requests to S.K.K. at the above address.
OTHER COMMENTS: This polymorphism is private for caucasians suggesting that it was generated after human racial divergence.
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