

### A three codon insertion/deletion polymorphism in the signal peptide region of the human apolipoprotein B (APOB) gene directly typed by the polymerase chain reaction

Eric Boerwinkle and Lawrence Chan<sup>1</sup>

Center for Demographic and Population Genetics, University of Texas Health Science Center at Houston, PO Box 20334, Houston, TX 77225 and <sup>1</sup>Departments of Cell Biology and Medicine, Baylor College of Medicine, Houston, TX 77030, USA

**SOURCE/DESCRIPTION:** Amplification of the first exon of the apo B gene was carried out by the polymerase chain reaction (PCR, ref 1) using the oligonucleotides 5'-CAGCTGGCGATGGACCCGCCGA-3' as the 5' primer and 5'-ACCGCCCTGGCGCCGCCAGCA-3' as the 3' primer.

**POLYMORPHISM:** The polymorphic alleles differ by the insertion or deletion of 9 bp and are directly visible after 8% polyacrylamide gel electrophoresis of the PCR products. The 9 bp encode amino acids -16 to -14 (Leu-Ala-Leu) of the apo B signal peptide (2).

**FREQUENCY:** Studied in 40 unrelated caucasians.

5'βIns - 0.575

5'βDel - 0.425

**CHROMOSOMAL LOCALISATION:** 2p23-24 (3).

**MENDELIAN INHERITANCE:** Codominant segregation was verified in 20 families.

**OTHER COMMENTS:** The denaturation step of the PCR was carried out at 94°C for 1 minute, and annealing and extension were carried out for 1.5 minutes at 64°C in the presence of 7% DMSO. The PCR product of the long allele is 93 bp and the short allele is 84 bp in length (figure). This polymorphism may have physiological significance because it involves the insertion/deletion of 3 amino acids in the signal peptide of a gene with a central role in lipid metabolism.

**REFERENCES:**

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