A unique length polymorphism in the signal peptide region of the apolipoprotein B gene in Mexican-Americans

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Source/Description: Amplification of the first exon of the apoB gene was carried out as previously described (1). The amplifying oligonucleotides were 5'-CAGCTGGCGATGGACCCGCCG-A-3' as the 5' primer and 5'-ACCGGCCCTGGCGCCCGCC-AGCA-3' as the 3' primer. Denaturation of the DNA was carried out at 94°C for 1 minute, annealing and extension were carried out for 1.5 minutes at 64°C. Ampification was carried out by 30 rounds of denaturation followed by resynthesis.

Polymorphism: In addition to the 2 apoB signal peptide alleles previously described for Northern European Caucasians (1), Mexican-Americans have yet another even longer apoB signal peptide allele. The three alleles, named for their lengths in amino acid residues, are visibly separated following electrophoresis in 8% polyacrylamide gels and ethidium bromide staining (figure).

Frequency: Studied in 451 Mexican-Americans from Starr County, Texas.
SPB-24: .345
SPB-27: .617
SPB-29: .029

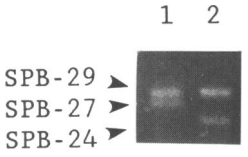
Chromosomal Localization: 2p23−24 (2).

Mendelian Inheritance: Verified codominant inheritance.

Other Comments: The SPB-29 allele has not been observed in any Caucasian sample investigated. This allele has, however, been observed in 2 other Amerindian populations and is likely diagnostic of Amerindian origins.

Acknowledgements: This work was supported by grants from the National Institutes of Health: HL-40613 to E.B., HL-34823 to C.L.H., and HL-27341 to L.C.

References: 1) Boerwinkle,E. and Chan,L. (1989) *Nucl. Acids Res.* **17**, 4003. 2) Chan,L. *et al.* (1985) *BBRC* **133**, 248.



A HindIII polymorphism in the human NCF2 gene

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Source/Description: A cDNA encoding one of the cytosolic factors (NCF2) deficient in a rare autosomal recessive form of Chronic Granulomatous Disease, $p67^{phox}$, was recently isolated by antibody screening of a stimulated HL-60 cell expression library (Leto *et al.*, 1990). Probe 67ES5' is a 0.84 kb fragment released by digestion of the clone with the restriction enzymes EcoRI and SacI, correspoding to position 1 to 843 of the cDNA.

Polymorphism: HindIII detects a simple two allele polymorphism with bands at either 3.8 kb (A1) or 2.1 kb (A2). There are also 2 constant bands at approximately 13 and 12 kb, the longer one detected when the entire cDNA is used as a probe.

Frequency: Estimated from 28 unrelated individuals:
A1: 0.80 ± 0.05
A2: 0.20 ± 0.05

Not Polymorphic For: AvaI, BamHI, EcoRI, KpnI, PstI, SacI, or XhoI in three heterozygotes tested.

Chromosomal Localization: The cDNA probe was used in Southern analysis of rodent×human somatic cell hybrid lines and chromosomal *in situ* hybridization to map NCF2 to band 1q25 (Francke *et al.*, 1990).

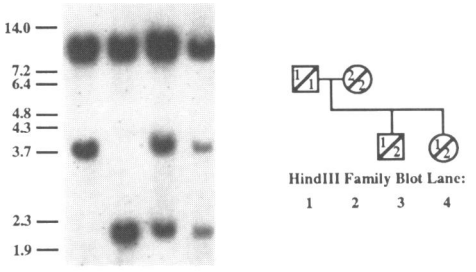
Mendelian Inheritance: Co-dominant segregation of the HindIII RFLP alleles was observed in two families, 10 individuals.

Probe Availability: The cDNA clone 10 encoding $p67^{phox}$ (GenBank # M32011) is available on request.

Other Comments: The RFLP pattern was observed under normal hybridization and washing conditions for a single copy probe.

Acknowledgements: We wish to thank Dr Neal Epstein for supplying DNA from several normal individuals.

References: 1) Leto *et al.* (1990) *Science* **248**, 727−730 2) Francke *et al.* (1990) *Am. J. Hum. Genet.* **47**, 483−492.



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