RFLPs for the human apolipoprotein B gene: HincII and PvuII

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SOURCE AND DESCRIPTION OF CLONE: 959 bp cDNA isolated from an adult human liver cDNA library. The clone contains the 5' coding region of apoB and 86 bp upstream of the initiation codon (Protter et al, 1986).

POLYMORPHISMS: HincII digestion gives four invariant bands of 1.8, 1.9, 4.0 and 5.4 kb, and a two allele polymorphism with bands at either 7.1 or 7.5 kb. PvuII digestion gives one invariant band of 2.5 kb and a two allele polymorphism of 5.6 or 8.1 kb.

FREQUENCIES: HincII (among 45 Europeans)	PvuII (among 44 Europeans)
7.5 kb allele: 0.88	8.1 kb allele: 0.92
7.1 kb allele: 0.12	5.6 kb allele: 0.08

NOT POLYMORPHIC FOR: PstI, HhaI, BglII, EcoRV (from 6 unrelated individuals), BclI, HinfI (from 8 ind.), KpnI (from 9 ind.), HindIII (from 10 ind.), XbaI (from 11 ind.), EcoRI (from 14 ind.) and MspI (from 16 ind.).

CHROMOSOMAL LOCALISATION: Probes from the 3'-end of the apoB gene have been assigned to 2p24 by in situ hybridization (Knott et al, 1985) and with somatic cell hybrids (Talmud et al, 1985).

MENDELIAN INHERITANCE: HincII: Segregation demonstrated in one family of 8 individuals. PvuII: Not investigated.

PROBE AVAILABILITY: Requests for probe to A. A. Potter at the above address.

OTHER COMMENTS: As calculated from 80 chromosomes there is linkage equilibrium between the two polymorphisms described here and between each of them and the XbaI polymorphism (Talmud et al, 1985).

REFERENCES: Knott, T.J. et al, (1985) Science 230, 37-43.

Protter, A.A. et al, (1986) Proc. Natl. Acad. Sci. USA 83, 1467-71.

Talmud, P.J. et al, (1985) Human Gene Mapping (Karger, ed) 8, 759.