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A RFLP associated with the low-density lipoprotein receptor gene (LDLR)

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SOURCE AND DESCRIPTION OF CLONE: pLDLR-2HH1, a 1.9kb fragment of the 3'end of the LDL receptor c-DNA clone (Yamamoto et al, 1984).

POLYMORPHISM: NcoI (CCATGG) identifies invariant bands of 9kb and 7kb, in addition to a simple two allele polymorphism with a band at either 3.4kb (N1) or 13kb (N2) (Figure).

FREQUENCY: Studied 52 normal Caucasians.
3.4kb allele (N1) 0.64
13kb allele (N2) 0.36

NOT POLYMORPHIC FOR: Eco R1, Bam HI, Hind III, Pst I, Taq I, Rsa I, Kpn I, Sca I, Cfo I, Bgl I, Ava I, Ban I, Ssp I, Nsi I, Sau I and Dde I with DNA from a panel of 5 unrelated Caucasians.

CHROMOSOMAL LOCALISATION: Gene localised to 19p13 (Naylor et al, 1985).

MENDELIAN INHERITANCE: Co-dominant segregation shown in 7 informative families (66 individuals).

REFERENCES: Yamamoto T, Davis CG, Brown MS, Schneider WJ, Casey ML, Goldstein JL and Russell DW. The human LDL receptor: a cysteine-rich protein with multiple Alu sequences in its mRNA. Cell 1984; 39: 27-38.
Naylor SL, Lalouel JM, Shaw DJ. Report of the committee on the genetic constitution of chromosomes 17, 18 and 19. Human gene mapping 8. Cytogenet Cell Genet 1985; 40: 242-246.

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