Three RFLPs upstream of the low density lipoprotein receptor (LDLR) gene

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SOURCE/DESCRIPTION: Cosmid RH15A spans a region of 40 kb of DNA immediately 5' to the LDLR gene.

POLYMORPHISMS: The whole cosmid was used as a probe. *Apa*I identifies a two-allele polymorphism (A1: 4.5 kb, A2: 4.2 kb), with several constant bands. *Bcl*I identifies a two-allele polymorphism (B1: 8.7 kb, B2: 5.5 kb), with several constant bands. A double-digestion with *Sac*I and *Kpn*I identifies a two-allele polymorphism (S1: 2.05 kb, S2: 2.0 kb), with several constant bands.

FREQUENCY: Estimated from 40 unrelated Caucasians;
A1: 0.04; A2: 0.96
Estimated from 18 unrelated Caucasians;
B1: 0.22; B2: 0.78
Estimated from 10 unrelated Caucasians;
S1: 0.50; S2: 0.50

NOT POLYMORPHIC FOR: *Bgl*I, *Bst*EII, *Bst*XI, *Dra*I, *Eco*RV, *Hinc*II, *Xmn*I, *Msp*I, *Taq*I, *Bam*HI, *Bgl*II, *Pst*I, *Eco*RI, *Hind*III. Also, single digestion with *Sac*I and *Kpn*I did not reveal polymorphism in those individuals who had polymorphic bands with the double digestion.

CHROMOSOMAL LOCALIZATION: LDLR gene has been mapped to 19p13.2.

MENDELIAN INHERITANCE: Co-dominant segregation of the alleles of each RFLP has been demonstrated in 3 two-generation families.

PROBE AVAILABILITY: Contact R.A.H.

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