BglII and EcoRI polymorphism of the human nm23-H1 gene (NME1)

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Source and Description: pNM23-H1 is a recombinant cDNA clone encoding the nm23-H1 human gene (1). The locus designation for this cDNA is NME1. The probe used is a 0.9 kb BamHI fragment of this clone.

BglII Polymorphism: BglII digestion identifies a two allele polymorphism with polymorphic bands at 2.3 kb and 7.6 kb as well as a constant band at 18 kb.

Frequency: As studied in 100 unrelated Caucasians.
2.3 kb allele (A1): 0.47
7.6 kb allele (A2): 0.53

EcoRI Polymorphism: EcoRI identifies a two allele polymorphism with polymorphic bands at 4.6 kb or 2.2 and 2.4 kb as well as constant bands at 21 kb, 1.7 kb.

Frequency: As studied in 100 unrelated Caucasians.
4.6 kb allele (B1): 0.58
2.2 and 2.4 kb allele (B2): 0.42
Observed frequencies fit Hardy-Weinberg equilibrium.

Not Polymorphic For: MspI, BamHI, HindIII, PstI, PvuII, RsaI, TaqI and XbaI.

Chromosomal Localization: Chromosome 17, p11-q11 using somatic cell hybrids and *in situ* hybridization (2).

Mendelian Inheritance: Co-dominant segregation shown in three informative two generation families.

Probe Availability: Request for probe to Patricia S. Steeg at Laboratory of Pathology, National Cancer Institute, NIH, Bethesda, MD 20892, USA.

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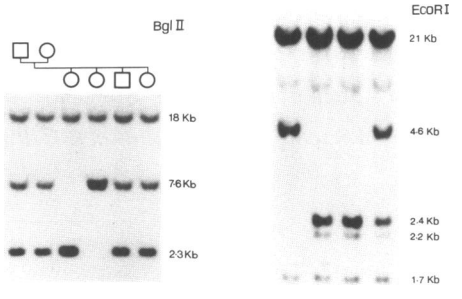

A *Hae*III polymorphism in the 3′ untranslated region of the low density lipoprotein receptor (LDLR) gene

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Description, Source and Method: The PCR primers SP11 and SP150 (1) amplify a 148 base pair fragment of the human low density lipoprotein receptor gene, which includes the 33 bp translated portion and 90 bp of the 3′ untranslated region of exon 18, as well as 25 bp of the preceeding intron.

Variation: From the published sequence (2), a *Hae*III site (GG△CC) is predicted to occur 56 bp downstream of the termination codon resulting, on digestion of the amplification product, in fragments of 114 bp and 34 bp. This site is polymorphic in the Caucasian population, as a result of a G to A transition (GACC), which destroys the *Hae*III site. The polymorphism was confirmed by sequencing both strands of both alleles in cloned PCR product from an individual heterozygous for the *Hae*III site. Several other polymorphisms have been reported in the 3′ untranslated region of the low density lipoprotein receptor gene (3, 4).

Frequency: Estimated in 24 unrelated Caucasian individuals by *Hae*III digestion of PCR products.

Pattern	Frequency
AA1 114/34 bp	0.77
AA2 148 bp	0.23

(Observed heterozygosity = .46).

Chromosome Localization: The LDLR gene has been assigned to chromosome 19p13.1-p13.3 (5).

Mendelian Inheritance: Codeminant inheritance demonstrated in one family (6 individuals).

Other Comments: Genomic DNA was used as a template for the polymerase chain reaction to amplify this region (6). Thirty cycles were performed, each of one minute denaturation at 95°C, one minute annealing at 50°C and one minute extension at 72°C. Buffer, nucleotide triphosphate and template concentrations were those recommended by the supplier of the *Taq* polymerase (Promega).

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