Three polymorphisms at the D17S29 locus

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Source/Description: A 4 kb *Eco*RI fragment from cosmid YNM67 was subcloned into the *Eco*RI site of pTZ19R.

Polymorphism: *Taq*I identifies a two allele polymorphism (A1: 3.4 kb, A2: 2 kb+ 1.3 kb) with constant bands of 0.86 kb and 1.45 kb. *Bgl*II identifies a two allele polymorphism (B1: 8.1 kb, B2: 6.7 kb) with no constant bands. *Bam*HI identifies a two allele polymorphism (C1: 5.1 kb, C2: 3.9 kb) with no constant bands.

Frequency: (estimated from the designated number (N) of unrelated Caucasians)

*Taq*I (N = 21)	*Bgl*II (N = 19)	*Bam*HI (N = 18)
A1: 0.45	B1: 0.21	C1: 0.78
A2: 0.55	B2: 0.79	C2: 0.22

Not Polymorphic For: *Rsa*I, *Pvu*II, *Hinf*I, or *Hin*dIII in a panel of 8 unrelated individuals. *Eco*RI identifies an apparent RFLP which is artifactual since it does not exhibit Mendelian inheritance.

Chromosomal Localization: pYNM67-R5 was assigned to chromosome 17p11.2 using a previously published chromosome 17-somatic cell hybrid panel (1, 2). This localization is based on the lack of hybridization to hybrid DH110-D1 retaining a del(17)(p11.2p11.2).

Mendelian Inheritance: Codominant segregation of these RFLPs has been observed in one large three-generation French-Acadian kindred (34 individuals).

Probe Availability: Contact ATCC.

Other Comments: Preassociation with human placental DNA is required prior to hybridization at normal stringency (65°C, 1 M Na^+) (2). *Taq*I and *Rsa*I RFLPs were previously detected by a 1.6 kb *Pst*I fragment from the cosmid YNM67 (Nakamura,Y., personal communication) but this probe often yielded high lane background.

Acknowledgements: We thank Drs. Yusuke Nakamura and Ray White for providing cYNM67. This work was supported by an MDA Task Force grant, by the Baylor Mental Retardation Research Center (HD24064-02), NIH grant R01 NS27042 and an Advanced Technology Program grant from the state of Texas to J.R.L and P.I.P.

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Tetranucleotide repeat polymorphism in the LPL gene

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Source/Description: A polymorphism $(TTTA)_n$ repeat is located in intron 6 of the lipoprotein lipase gene at the 3′ end of an Alu sequence (1). The polymerase chain reaction (PCR) was used to selectively amplify the sequence from genomic DNA, using two oligonucleotides flanking the repeat. The expected size of the fragment is 127 bp.

Primer Sequence:
GZ–14 = ATCTGACCAAGGATAGTGGGATATA
GZ–15 = CCTGGGTAACTGAGCGAGACTGTGTC

Frequency: estimated in 20 unrelated Caucasian American individuals:

Allele (nt)	Number of (TTTA) Repeats	Frequency
131	12	0.08
127	11	0.46
123	10	0.46

Heterozygosity index is 54% and the alleles are in Hardy-Weinberg equilibrium.

Mendelian Inheritance: Co-dominant segregation was observed in one family with 12 informative meiosis.

Chromosomal Localization: LPL gene has been assigned to chromosome 8p22 (2).

Other Comments: The PCR was performed as previously described (3) using end-labeled oligo GZ-14 and unlabeled oligo GZ-15 with the following modifications: 1) denaturation at 96°C for 1 min., 2) annealing and extension at 68°C for 3 min., and 3) the number of cycles was 25. PCR product was fractionated on 8% denaturing polyacrylamide gel. The size of allele was determined by comparison with end-labeled MspI digested pBR322 DNA (Figure).

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