BclI and MspI polymorphisms at the D6S90 locus

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Source/Description: pHZ-71 is a 2.8 kb fragment with *EcoRI* ends cloned in pBluescript II KS. It was subcloned from λHZ-71 which was isolated from a genomic library constructed in λDASH from a somatic cell hybrid retaining chromosome 6p as the only human chromosome (1).

Polymorphism: *BclI* identifies a two allele polymorphism: 2.7 kb (B1) and 2.2 kb (B2). A 6.0 kb constant band was present. A two allele polymorphism was also identified using *MspI* in U.S. Blacks. The fragment sizes were 2.55 kb (M1) and 2.3 kb (M2). A 4.3 kb constant band was present.

Frequency:

	A1	A2	B1	B2
80 unrelated Caucasians	0.8	0.2		
9 unrelated U.S. Blacks	0.61	0.39	0.28	0.72

Not Polymorphic For: *ApaI*, *BamHI*, *BanI*, *BanII*, *BglI*, *BglII*, *DraI*, *EcoRI*, *EcoRV*, *Fnu4HI*, *HaeIII*, *HindIII*, *HinfII*, *MboI*, *PstI*, *PvuII*, *RsaI*, *ScaI*, *SphI*, *SstI*, *TaqI*, *XbaI*.

Chromosomal Localization: pHZ-71 localizes to 6p11–6p21 using a previously published chromosome 6 somatic cell hybrid panel (1, 2). Pairwise linkage analysis between pHZ-71 and two chromosome 6p markers, D6S4 and D6S19 in CEPH families gave a maximum lod score of 15.4 at theta = 0.07 with D6S4, and a lod score of 6.5 at theta = 0.12 with D6S19 confirming the localization to proximal 6p. Mendelian inheritance was observed in all cases.

Probe Availability: Contact H.Y.Zoghbi.

Other Comments: Prehybridization with human placental DNA is required prior to hybridization at normal stringency (65°C 1 M Na⁺).

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References: 1) Zoghbi *et al.* (1990) *Genomics* 6, 352. 2) Zoghbi *et al.* (1988) *Genomics* 3, 396.

Sequence polymorphism in the human apoB gene at position 8344

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Source/Description: ApoB exon 26 clones were obtained by polymerase chain reaction (PCR) from genomic DNA of one individual. Sequencing revealed heterozygosity in nucleotide 8344. A substitution of C to T resulted in a proline (CCA) to leucine (CTA) change in amino acid 2712. This residue is in a proline-rich cluster and may be involved in a β-turn structure (1). Disruption of proline in this region might affect apoB function.

Primer Sequence: The 5' primer used for PCR was: AAGAAGCTTAATGAATTATCTTT; the 3' primer used for PCR was TGGGGAGCTCGTCGTGAGATT. The reaction was carried out in a DNA thermal cycler for 40 cycles of: 1 min at 92°C, 2 min at 55°C and 3 min at 65°C. The concentration of reagents used was as recommended by the manufacturer of Taq polymerase (Perkin-Elmer Cetus).

Frequency: Estimated from 58 unrelated Caucasians by allele-specific hybridization (ASO).

Proline allele: 83%

Leucine allele: 17%

Chromosomal Location: The apoB gene has been assigned to chromosome 2p23–24 (2).

Mendelian Inheritance: Co-dominant segregation was observed in 5 two or three generation families.

Other Comments: Sequences used for ASO probes are: TCACATACCAGAATTCC for proline allele; TCACATACTAGAATTCC for leucine allele. ApoB sequences used were reported previously (3).

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