A TaqI RFLP detected by the human haptoglobin (HP) cDNA probe, pULB1148

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SOURCE/DESCRIPTION: pULB1148 has a 1.4 kb HP cDNA insert in the PstI site of pBR322 (Straten, A. et al., 1983).

POLYMORPHISM: TaqI (New England Biolabs) identifies an invariant band of 7.0 kb and a simple two allele polymorphism with a band of either 7.2 kb (F.1) or 5.8 kb (F.2).

FREQUENCY: TaqI studied in 55 Australian individuals.
7.2 kb allele (F.1) 0.58  5.8 kb allele (F.2) 0.42

CHROMOSOMAL LOCALATION: The haptoglobin gene has been localised to 16q22.105->16q22.108 by Southern blot analysis of human-mouse cell hybrids (Callen, D.F. et al., 1988).

MENDELIAN INHERITANCE: Co-dominant inheritance was shown in the three generation Utah pedigree K-1331. The three generation family is shown below.

PROBE AVAILABILITY: Available from A. Bollen, Université Libre de Bruxelles, Service de Génétique Appliquée, rue de l'industrie 24, B-1400 Nivelles, Belgium.

OTHER COMMENTS: The following haplotype frequencies were observed among 25 unrelated Utah individuals: C.1/F.1 0.14, C.1/F.2 0.4 and C.2/F.1 0.46. No C.2/F.2 haplotype was detected. EcoRI identifies the 10.1 kb (C.1) allele and the 11.8 kb (C.2) allele (Oliviero, S. et al., 1985). A significant non-random allelic association (linkage disequilibrium) was detected for C and F alleles (D = -0.18; $X^2_1 = 25.4$).

REFERENCES: Callen, D.F. et al. (1988), Genomics (In Press). Oliviero S., et al. (1985), Hum. Genet. 70: 66-70. Straten, A. et al. (1983), EMBO J. 2: 1003-1007.

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