MspI RFLP at CRYB1 locus (17q11.2 – 17q12)

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SOURCE/DESCRIPTION: The probe pbeta8-2 is a 1.7 kb MspI fragment cloned into the AccI site of pUC8. The fragment was derived from the clone Lambda18A (1).

POLYMORPHISM: MspI (CCGG) identifies two alleles with lengths of 2.4 and 1.7 kb respectively. No constant bands are present.

FREQUENCY: In 145 unrelated individuals.

2.4 kb allele	.41
1.7 kb allele	.59

NOT POLYMORPHIC FOR: BglII, PstI, HindIII, TaqI and RsaI tested with five individuals. The parental clone Lambda18A also shows no polymorphism with AatI, BanII, DraI, PvuII, ScaI, StyI or EcoRI.

CHROMOSOME LOCALIZATION: CRYB1 has been assigned to 17q11.2->17q12 (2). The localization of pbeta8-2 to this region has been confirmed by genetic linkage studies which show this probe to be a member of a group showing tight linkage to NF1 and each other.

MENDELIAN INHERITANCE: No significant deviation from Hardy-Weinberg equilibrium was found. Proper Mendelian segregation was observed in 125 offspring of backcross matings and 68 offspring of intercross matings.

PROBE AVAILABILITY: Submitted to the ATCC.

OTHER COMMENTS: pbeta8-2 is single copy, requiring no probe prehybridization with human placental DNA. A PstI polymorphism associated with Lambda8A (1), another putative CRYB1 clone, has been found not to be linked to other chromosome 17q markers, indicating an homology of that clone with sequences not on chromosome 17q.

REFERENCES:

1. Hogg et al. (1986) J Biol Chem **261**, 12420-12427.
2. vanTuinen et al. (1987) Genomics **1**, 374-381.

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