A rare PvuII RFLP at the CRYB1 locus (17q11.2 – 17q12)

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SOURCE/DESCRIPTION: The probe LambdalbetallA has been described by Hogg et al. (1).

POLYMORPHISM: PvuII (CAGCTG) identifies two alleles with lengths of 5.6 kb and 4.6 kb. Constant fragments are 3.6 kb, 2.0 kb, 1.9 kb and 1.2 kb.

FREQUENCY: In 50 unrelated individuals:

5.6 kb allele	.97
4.6 kb allele	.03

NOT POLYMORPHIC FOR: BglII, PstI, HindIII, TaqI, AatI, BanII, DraI, StyI, EcoRI and RsaI when tested with five individuals.

CHROMOSOME LOCALIZATION: CRYB1 has been assigned to 17q11.2->17q12(2). The localization of LambdalbetallA to this region has been confirmed by physical localization studies with chromosome 17 deletion and translocation hybrids (unpublished results).

MENDELIAN INHERITANCE: No deviation from Mendelian segregation was observed in offspring of two families with heterozygous parents.

PROBE AVAILABILITY: Submitted to the ATCC.

OTHER COMMENTS: LambdalbetallA requires probe prehybridization with human placental DNA. Stringencies of hybridization and post hybridization washes are otherwise normal. The enzyme ScaI (AGTACT) may also reveal a polymorphism in disequilibrium with the PvuII site polymorphism.

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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