A moderately frequent RFLP identified by both SacI and BanII with a probe from the HOX2 locus in man (17q11-17q22)

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SOURCE/DESCRIPTION: Subclone BS3, a 2.6 kb EcoRI/SacI fragment in bluescript M13 (Stratagene), from a genomic clone, λ 86x5B (14 kb EcoRI insert in Charon 4A), containing two homeoboxes, HU-1 and HU-2 (Levine et al., 1984). The subclone, BS3, is approximately 5 kb 5' to HU-2.

POLYMORPHISM: SacI (GAGCT/C) and BanII (GPuGCPy/C) (New England Biolabs) identify the same polymorphism. The pattern with SacI is one invariant band at 1.9 kb and a simple codominant two allele polymorphism with either a band at 2.0 kb or bands at 1.1 kb and 0.9 kb. The BanII pattern has the same invariant band at 1.9 kb and polymorphic bands either at 1.7 kb or at 0.9 kb and 0.8 kb.

FREQUENCY: Studied 45 unrelated European Caucasians:

SacI 2.0 kb : (A1) = 0.85 $\pm$.01
BanII 1.7 kb

SacI 1.1 kb and 0.9 kb : (A2) = 0.15 $\pm$.01
BanII 0.9 kb and 0.8 kb

NOT POLYMORPHIC FOR: BamHI, BglII, BstEII, BstNI, DraI, EcoRV, HindIII, HincII, KpnI, PstI, PvuII, TaqI, and XmnI, using a panel of 10-12 unrelated Caucasians.

CHROMOSOMAL LOCALISATION: 17q11-q22 (Joyner et al., 1985; Rabin et al., 1985)

MENDELIAN INHERITANCE: Autosomal codominant segregation observed in four large reference pedigrees with 15 informative matings.

PROBE AVAILABILITY: Available for collaboration from F.H. Ruddle, Department of Biology, Yale University, New Haven, CT 06510 USA.

OTHER COMMENTS: An infrequent polymorphism for RsaI was noted in two families.

REFERENCES:

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