An anonymous single copy genomic clone at 13q12-13q13 identifies three RFLPs [HGM8 assignment no. D13S11]

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SOURCE AND DESCRIPTION OF CLONE: pG2E3.1, a 3.1 kb EcoRI fragment subcloned into pBR329 from the 14.3 kb genomic insert of phage clone 13G2, isolated from a Sau3AI library (vector EMBL4) containing chromosome 13 in duplicate as its only human genetic material (Scheffer et al., 1985).

POLYMORPHISMS: MspI (C/CGG)(Boehringer Mannheim) identifies a simple two-allele polymorphism with bands at either 5.4 kb (A1) or 4.4 kb (A2). PstI (CTGCA/G)(Boehringer Mannheim) identifies one invariant band at 12.8 kb and a simple two-allele polymorphism with bands at either 7.6 kb (B1) or 6.7 kb and 0.9 kb (B2). HpaI (GTT/AAC)(Anglian Biotechnology Ltd.) identifies a simple two-allele polymorphism with bands at either 15.0 kb (C1) or 13.0 kb (C2).

FREQUENCY: MspI: Studied 23 Caucasians, 5.4 kb allele (Al): 0.65, 4.4 kb allele (A2): 0.35. PstI: Studied 21 Caucasians out of the 23 above, 7.6 kb allele (B1): 0.33, 6.7/0.9 kb allele (B2): 0.67. HpaI: Studied a different group of 10 Caucasians, 15.0 kb allele (C1): 0.80, 17.0 kb allele (C2): 0.20.

NOT POLYMORPHIC FOR EcoRI, Bg1II, HindIII, TaqI, SinI, BstEII, PvuII and BamHI with DNA from 10 unrelated Caucasians.

CHROMOSOMAL LOCALISATION: Probe localised to 13q12-13q13 using panel of somatic cell hybrids (Buys et al., 1985).

MENDELIAN INHERITANCE: Co-dominant segregation shown in 3 informative families, 34 individuals.

PROBE AVAILABILITY: Available for collaboration. Contact C.H.C.M. Buys.

OTHER COMMENTS: For digestion to completion overnight incubation with MspI (2 units/microgram genomic DNA) should be followed by two additional 2 h incubations with 2 units/microgram DNA. This is not necessary with PstI and HpaI. The MspI and PstI RFLPs show strong linkage disequilibrium.

REFERENCE: Scheffer et al., (HGM8) Cytogenet. Cell Genet. $\underline{40}$ (1985)738; Buys et al., ibid. (1985)597.

 ${\tt ACKNOWLEDGEMENTS:}$ This work was supported by the Netherlands Cancer Foundation.