Three RFLPs recognized by an anonymous sequence localized to 21q11.2 [HGM8 D21S72]

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SOURCE AND DESCRIPTION OF CLONE: Three unique HindIII/EcoRI sequences were subcloned into pUC8 from a single lambda clone isolated from the G95α1 library (partial MboI digested DNA ligated into EMBL 3B) obtained from J. Gusella using DNA from a 20/X human/hamster hybrid cell line (Bruns et al, 1982) that had been selected with 6 thioguanine to eliminate the X chromosome. Subclones were designated pG95α1-11a,-b and -c and are 1.50, 0.45 and 0.55 kb respectively.

POLYMORPHISM: Subclones -a, -b and -c detect TaqI RFLPs designated A1 (3.95 with the -a subclone or 1.60 kb with the -b or -c subclones) and A2 (5.55 kb with all three subclones). The -a subclone detects the A alleles in EcoRI as well as TaqI digested DNAs (EcoRI A1 = 1.95 kb, A2 = 2.95 kb). Subclones -b and -c detect HincII alleles designated B1 (2.60 kb) and B2 (2.30 kb). Subclone -c detects alleles C1 (1.95 kb) and C2 (7.75 kb) with a constant band of 0.4 kb in PvuII digested DNAs.

FREQUENCY: A and B alleles were studied in 34 and the C in 20 random placentae (most were likely Caucasian). A1 (3.95 or 1.60 kb) = 0.68; A2 (5.55 kb) = 0.32; B1 (2.60 kb) = 0.69; B2 (2.30 kb) = 0.31; C1 (1.95 kb) = 0.55; C2 (7.75 kb) = 0.45.

NOT POLYMORPHIC FOR: Subclone a--MspI (34), BglI (30), HaeIII (34), XbaI (34) HindIII (34), BamHI (30), BstEII (30), KpnI (30), CfoI (30), ClaII (4), PstI (4), SstI (4), subclone b--MspI (30), subclone c--MspI through CfoI as for subclone a in 30 unrelated chromosomes. Numbers in brackets = unrelated chromosomes tested.

CHROMOSOMAL LOCALIZATION: pG95α1-11a subclone was localized to 21q11.2 using in situ hybridization.

MENDELIAN INHERITANCE: Co-dominant segregation was shown for the A alleles in 108 members of four kindreds and in 55 members of three kindreds for the B alleles.

PROBE AVAILABILITY: Freely available.

REFERENCE: Bruns et al, Cytogenet Cell Genet 32:256, 1982.

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