BglII and EcoRV RFLPs on the Human DQB locus

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Source/Description: 1426 bp full cDNA probe cloned into pUC12. Insert is excised by BamHI digestion (1).

Polymorphism: EcoRV identifies a two allele polymorphism with bands at 4.3 and 3.0 kb plus one invariant band at 13.0 kb. BglII identifies a fragment of 11.0 kb corresponding to the 4.3 EcoRV band. The 3.0 EcoRV fragment appears as a BglII fragment of either 9.2 or of 8.8 kb. A non-polymorphic band of 6.4 kb is always present. In every lane examined always two of the three fragments were present suggesting the existence of three alleles for Dobeta.

Frequency: Frequency estimated in 9 caucasians and 6 mixed caucasian/negro, indian or oriental individuals (São Paulo population): 11.0 kb fragment = 0.2; 8.8 kb fragment = 0.6 and 9.2 kb fragment = 0.2.

Not Polymorphic For: TaqI, PvuII, HindIII, PstI.

Chromosomal Localization: Localized on the HLA region of the short arm of chromosome 6 (between DQB2 and DPA1).

Mendelian Inheritance: Co-dominant inheritance as other HLA genes, segregates with HLA class II haplotypes in 2 families.

Probe Availability: Commercialized by Appligene.

Reference: 1) Tonnelle *et al.* (1985) *EMBO J.* **4**, 2839–2847.

Figure: Dobeta polymorphic fragments as observed with BglII digests of homozygous typing cells.

A TaqI polymorphism identified by 26-6 (D16S125) proximal to the locus affecting adult polycystic kidney disease (PKD1) on chromosome 16

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Source/Description: Probe 26-6 (D16S125) is a 0.5 kb fragment subcloned in pKUN1 (1) after PstI digestion of a cosmid (218) from a random MboI library of a single human chromosome 16 hybrid cell line (CY18) (2).

Polymorphism: The probe detects a two-allele restriction site TaqI polymorphism with fragment sizes 5.8 kb (E1) and 0.85 kb (E2). It also detects two constant bands, one of 0.65 kb (Ec) and one of 0.2 kb (Ec). Polymorphism was also demonstrated with EcoRI.

Not Polymorphic For: HindIII, MboI, PstI, PvuII, RsaI, SacI and XmnI.

Frequency: The allele frequencies were studied in 55 unrelated individuals. Fragment 1 (E1) 0.30 and fragment 2 (E2) 0.70.

Chromosomal Localization: The probe is localized proximal to the locus affecting PKD1 and distal to breakpoint GM2324 t(16;22) (p13.3;q21) (3).

Mendelian Inheritance: The polymorphism segregates in a Mendelian fashion in a large number of families with PKD1.

Probe Availability: The probe is available for diagnostic purposes.

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References: 1) R.N.H.Konings *et al.* (1986) *Gene* **46**, 269–276. 2) D.F.Callen (1986) *Ann. Genet.* **29**, 235–239. 3) M.H.Breuning *et al.* (1989) *Cytogenet. Cell Genet.* **51**, 969.