Polymorphism at the D20S13 (p ϕ 64) locus

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Source and Description: The probe was isolated from a Charon 21A bacteriophage human chromosome 20 specific library (1), and subcloned as a HindIII fragment into pUC19.

Polymorphism: PvuII detects a two allele polymorphism with allelic bands at either 4.4 kb or 3.4 kb. MspI detects a three allele polymorphism with allelic bands at 9.4 kb, 8.2 kb or 6.3 kb. No constant bands appear with PvuII but a faint constant band occurs at 4.2 kb with MspI.

Frequency: Gene frequencies are based on a sample of 278 chromosomes from 139 unrelated persons of mixed Caucasian ancestry.

	Allele	Frequency	Allele	Frequency
PvuII	A1 4.4 kb	0.78	MspI B1 9.4 kb	0.78
	A2 3.4 kb	0.22	B2	8.2 kb
			B3 6.3 kb	rare

Except when a rare allele occurred with MspI in 5 related individuals in one family, PvuII and MspI show complete linkage disequilibrium ($A1B1 = .78$; $A2B2 = .22$) suggesting a single difference in the DNA may be causing both RFLPs, for example, a small insertion/deletion.

Not Polymorphic For: RsaI, DraI, StuI, EcoRI, SacI, BamHI, BglII, HindIII, PstI on a panel of 11 unrelated persons.

Chromosomal Localization: Using a somatic cell hybrid panel (2), the probe was localized to chromosome 20p. Preliminary linkage analyses (3) give lod scores above 4 with D20S5 and D20S6, confirming the mapping to 20p.

Mendelian Inheritance: Mendelian inheritance has been demonstrated in 7 large families.

Probe Availability: Freely available from R.W.Deed or K.K.Kidd.

Other Comments: p ϕ 64 has a repeat element and thus requires hybridization with an excess of unlabelled denatured DNA.

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References: 1) van Dilla, M.A. *et al.* (1986) *Biotechnology* **4**, 537–552. 2) Sacchi, N. *et al.* (1986) *Science* **231**, 379–382. 3) Chappell, P.B. *et al.* (1989) *Cytogenetics and Cell Genetics* **51**, 975–976.

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Two EcoRI RFLPs at the GLUT2 locus

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Source and Description: pGEM 4Z/HTL 3 is a human glucose transport-like protein (GLUT2) cDNA clone containing 2.4 Kb SalI insert.

Polymorphism: Restriction endonuclease EcoRI identifies two polymorphisms. The first is a two allele polymorphism (E1), with bands at 5.6 Kb and 3.4 Kb. The second is a two allele polymorphism (E2) with bands at 1.9 Kb and 1.8 Kb. The invariant bands are at 6.8 Kb, 4.5 Kb and 3.8 Kb.

Frequencies: Studied in 22 unrelated European Caucasian individuals:

E1 — 5.6 Kb allele: 0.84	E2 — 1.9 Kb allele: 0.07
3.4 Kb allele: 0.16	1.8 Kb allele: 0.93

Chromosomal Localization: Assigned to chromosome 3q26.1–3q26.3 using a panel of mouse-human somatic cell hybrids and by in situ hybridization to metaphase chromosomes (1).

Mendelian Inheritance: Co-dominant segregation of the two polymorphisms (E1 and E2) observed in one Caucasian family (19 individuals).

Probe Availability: Contact G.I.Bell.

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Reference: 1) Fukumoto, H. *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 5434–5438.