An EcoRI RFLP in the human $\alpha 2$ (XI) (COL 11A2) gene

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Source/description of clone: pSC2.1 is a BamHI-EcoRI 4.5kb subclone of the cosmid clone cosHcol.11 containing part of the human 2(XI) collagen (COL11A2) gene (1). pSC2 does not cover the polymorphic EcoRI site but the 6.0 kb XhoI subclone, pVL19, does.

Polymorphism: Using pSC2.1 as a probe, a 2 allele polymorphism is identified (A1:11.3kb; A2:8.6kb). Using a probe (pVL19) which covers the polymorphic site, a third fragment of 2.5kb derived from cleavage of allele A1 is detected.

Frequency: 49 unrelated Chinese allele A1:0.79 A2 0.21
41 unrelated English caucasians allele A1: 0.59 A2 0.41

Not polymorphic for: TaqI, RsaI (100 chromosomes tested).

Chromosomal localisation: COL11A2 has been mapped to chromosome 6p2.12 and is close to the HLA locus (1).

Mendelian inheritance: Autosomal codominant. 17 meioses tested in one family.

Probe availability: Contact Dr. Cheah.

Other comments: Probe does not contain repetitive sequences.

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Reference: (1) Kimura,T., Cheah,K.S.E., Chan,S.D.H., Lui,V.C.H., Matthei,M.-G., van der Rest,M., Ono,K., Solomon,E., Minomiya,Y. and Olsen,B.R. (1989) *J. Biol. Chem.* in press.

RFLP for human DBH (dopamine beta-hydroxylase)

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Source/description: 1.9 kb single copy fragment amplified by PCR from human genomic sequences in exons 10 and 12, including exon 11 (Lamoureux *et al.*, 1987, Kobayashi *et al.*, 1980).

Polymorphism: XbaI digestion gives a two allele polymorphism with bands at 10.0 kb and 9.6 kb.

Frequency: Among 34 unrelated North Americans
10.0 kb allele (A1) .15
9.6 kb allele (A2) .85

Not polymorphic for: EcoRI, BamHI, HindIII, BglII, PstI, SacI, BglII, XmnI, DraI, HincII, EcoRV, StuI, BclI, BanI, StyI, RsaI, TaqI, MspI.

Chromosomal localisation: The probe has been localized to chromosome 9q34 by *in situ* hybridization (Craig *et al.*, 1988).

Mendelian inheritance: Codominant segregation pattern has been observed in 19 families (196 individuals).

Probe availability: The probe and primers are freely available from C. Craft.

Other comments: The sense primer is derived from nucleotides 1525–1554 and the antisense primer from nucleotides 1807–1848 of the published cDNA seg. (Lamoureux *et al.*, 1987).

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References: Craig *et al.* (1988) *Cytogenet. Cell Genet.* **48**, 48–50. Lamoureux *et al.* (1987) *EMBO J.* **6**, 3931–3937. Kobayashi *et al.* (1989) *Nucl. Acids Res.* **17**, 1089–1102.