Sacl polymorphism at the human TCR delta chain constant region (TCRD)

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Source and Description of Clone: pGTC2&, a 700 bp human TCR Delta Chain Constant Region cloned into EcoRI site of pGEM2.

Polymorphism: SacI identifies two-allele-RFLP with bands at 16 kb (A1) and 11 kb (A2). The genomic DNA source was peripheral blood leucocytes.

Frequency: Estimated from 52 Spanish individuals: A1 (16 kb): 0.32 A2 (11 kb): 0.68 PIC = 0.34

Not Polymorphic for: EcoRI, RsaI, HindIII, TaqI, BamHI, BgIII, PvuII and PstI.

Chromosomal Localization: Chromosome 14q11.2 (1, 2).

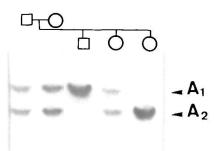
Mendelian Inheritance: Codominant in four 2 generation families with 18 members.

Probe Availability: Dr. Peter van den Elsen, Department of Immunohaematology and Bloodbank, Leiden, The Netherlands.

Other Comments: Standard hybridization and washing procedures.

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References: 1) Hata, S. et al. (1987) Science 238, 678. 2) Isobe, M. et al. (1988) PNAS 85, 3933-3937.



Mspl RFLP at the D5S122 locus tightly linked to APC

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Source and Description: pCB83.6 is a 300 bp PstI fragment isolated from cosmid cCB83 (D5S122), and subcloned in pKUN (1). This cosmid was isolated from a cosmid library which was generated from a reduced complexity radiation hybrid containing fragments of human chromosome 4 and 5 in a Chinese hamster background (2).

Polymorphism: MspI identifies a three allelic polymorphism with fragments of 10.0 kb (allele 1), 8.5 kb (allele 2) and 7.5 kb (allele 3). No constant bands are detected (see Figure 1).

Frequency: Studied in 46 unrelated European Caucasian individuals.

Allele 1 = .02 Allele 2 = .43 Allele 3 = .55

Not Polymorphic For: EcoRI, HindIII, PstI and TaqI (14 chromosomes tested).

Chromosomal Location: Assigned to the long arm of chromosome 5 (q15-q23) using a panel of reduced complexity human-Chinese hamster radiation hybrids (1) and by fluorescence in situ hybridization to human lymphocyte metaphase chromosomes (3).

DNA Digestion and Hybridisation Conditions: MspI digestions were done overnight at room temperature in the buffer recommended by the supplier. All hybridisations were performed at 42°C in 40% formamide, 0.1% SDS, 5 mM EDTA, 0.9 M NaCl, 50 mM Na₂H₂PO₄ pH 7.4, 0.1 mg/ml sonicated salmon sperm DNA, $5 \times$ Denhardts and 10% dextran sulphate.

Mendelian Inheritance: Co-dominant segregation was observed in 6 Dutch adenomatous polyposis coli families. Linkage analysis in those families revealed no recombinant between *APC* and D5S122 in 39 informative phase known meioses, giving a maximum lod score of 8.63 at theta 0.00.

Probe Availability: Contact C.Breukel or P.Meera Khan.

Acknowledgement: We would like to thank H.Dauwerse for the *in situ* hybridisation.

References: 1) Konings, R.N.H. et al. (1986) Gene 46, 269–276. 2) Tops et al. in preparation. 3) Kievits, et al. (1990) Cytogenetics and Cell Genetics 53, 134–136.

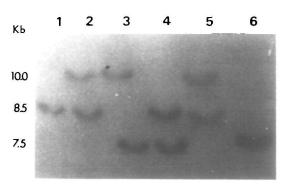


Figure 1. Autoradiogram of a Southern blot with human DNA samples digested with MspI, after hybridisation with pCB3.6. Phenotypes: lane 1: 2-2, 2:1-2, 3: 1-3, 4: 2-3, 5: 1-2 and 6: 3-3.