

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

E.Martínez-Naves, E.Coto, J.M.Urra, F.Setién and C.Lopez-Larrea

Servicio de Inmunología, Hospital Covadonga, 33006 Oviedo, Spain

Source and Description of Clone: pGTC2δc, 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, BglII, PvuII and PstI.

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

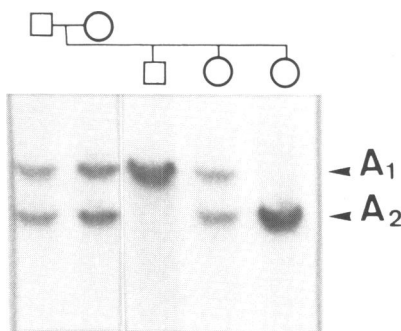
Mendelian Inheritance: Codominant in four 2 generation families with 18 members.

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

Other Comments: Standard hybridization and washing procedures.

Acknowledgements: We thank Dr. P. van den Elsen for providing the TCR delta chain probe. This work was supported by a grant FISS 89/0370.

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



MspI RFLP at the D5S122 locus tightly linked to APC

C.Breukel, C.Tops, E.Ras and P.Meera Khan

Department of Human Genetics, University of Leiden, The Netherlands

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×Denhardtts 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.

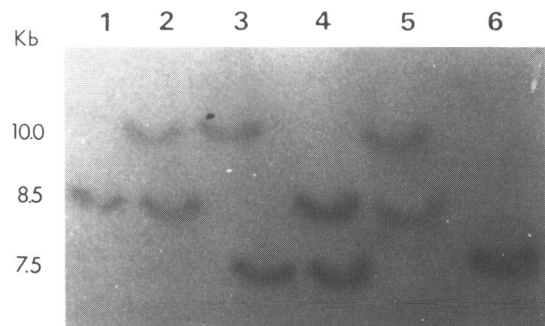


Figure 1. Autoradiogram of a Southern blot with human DNA samples digested with MspI, after hybridisation with pCB3.6. Phenotypes: lane 1: 2–2, 2: 1–2, 3: 1–3, 4: 2–3, 5: 1–2 and 6: 3–3.