

## A BglII RFLP detected by the probe JA8-1 at human chromosome band 11p13 (D11S417)

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**Source and Description of Probe:** The probe JA8-1 BE0.6 is a 600 bp single-copy BamHI/EcoRI fragment derived from a  $\lambda$  phage clone.  $\lambda$ JA8-1 was obtained by screening a human chromosome jumping library (1) with the probe A8 (2). The sequence was cloned into the pUC19 plasmid at BamHI and EcoRI sites to yield the clone pJA8-1 BE0.6 which was transformed into DH5 $\alpha$  *E. coli* cells.

**Polymorphism:** BglII (AGATCT) (Boehringer-Mannheim) detects a seven allele polymorphism with an invariant fragment of 7.4 kb and polymorphic fragments of 2.28, 2.21, 2.07, 2.00, 1.93, 1.79, or 1.72 kb. PstI also detects this apparent deletion/insertion polymorphism, but with less resolution.

**Allele Frequency:** Ascertained in 11 unrelated individuals.

2.28 kb allele: 0.05  
2.21 kb allele: 0.05  
2.07 kb allele: 0.09  
2.00 kb allele: 0.18  
1.93 kb allele: 0.09  
1.79 kb allele: 0.45  
1.72 kb allele: 0.09

**Not Polymorphic For:** HindIII, EcoRI

**Chromosomal Localization:** The probe JA8-1 maps proximal of D11S87 on the same 500 kb NotI restriction fragment in chromosomal band 11p13 (2), as shown by pulsed field-gradient electrophoresis.

**Mendelian Inheritance:** Co-dominant segregation observed in three families (31 individuals).

**Probe Availability:** Contact Dr. Grady F. Saunders.

**References:** 1) Collins, F.S. and Weissman, S.M. (1984) *Proc. Natl. Acad. Sci. USA* **81**, 6812–6816. 2) Compton, D.A., Weil, M.M., Yeger, H. *et al.* (1989) *Genomics* in press.

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## Two novel HLA-DRB Taq I RFLPs distinguish HLA-Bw57-DR7-DRB4-Null from DRB4-positive haplotypes

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**Source and Description of Clone:** A 517 bp PstI fragment of an HLA-DRB clone, pRTV1 probe (1), was used.

**Polymorphism:** Genomic DNA Taq I digests hybridized with the DRB probe show two novel RFLPs of 3.8 and 3.1 kb.

**Frequency of Presence:** Analyzed unrelated individuals, n=194; positive for the 3.8 kb band, n=81 (42%); positive for the 3.1 kb band, n=6 (3%). The 3.8 kb RFLP is only present in all haplotypes bearing HLA-DR4, 9,7.1 and 7.2a\* (DQw2); the 3.1 kb RFLP is only present in all haplotypes bearing HLA-DR7.2b\* (DQw9).

**Not Polymorphic For:** Bgl II, Sst I, Hind III and Eco RI.

**Chromosomal Localization:** HLA region on chromosome 6.

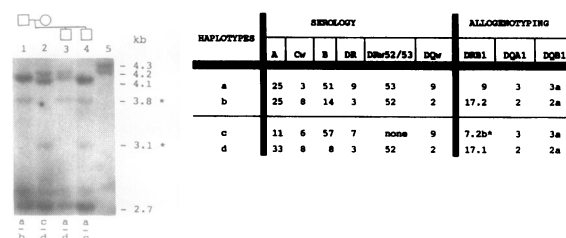
**Mendelian Inheritance:** Demonstrated in 12 families (33 sibs).

**Probe Availability:** Dr. J.L.Bidwell, United Transplant Service, Southmead Road, Bristol BS10 5ND, UK.

**Other Comments:** a) DR7-DQw9 haplotypes may now be distinguished from DR9-DQw9 haplotypes using pRTV1 DNA hybridization of Taq I digests. b) The 3.1 kb RFLP may thus be used as a novel marker of the highly conserved HLA-B57-DR7-DQw9-BFS-C2C-C4A6-C4B1 extended haplotype, present in five out of the six 3.1 kb +ve individuals. c) These novel RFLPs identify DRB4-bearing haplotypes which do (3.8 kb +ve, 3.1 kb -ve; DR4,7.1,7.2a\*,9) or do not (3.8 kb -ve, 3.1 kb +ve; DR7.2b\*) express the DR $\beta$ IV protein. This gene product (DRw53) is not expressed in HLA-DR7-DQw9 haplotypes (DRB4-null) due to an altered splicing site in the DRB4 gene (2).

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**References:** 1) Bidwell. *et al. Mol. Immunol.* **23**, 1111–1116 (1986), 2) Sutton *et al. Immunogenetics* **29**, 317–322 (1989).



Lane 5: Unrelated individual not carrying the DRB4 gene (HLA-DR5, 6). \*: Novel HLA-DRB RFLP alleles.