DdeI polymorphism in the HLA-DRA regulatory region

J.F.Eliaou, V.Pinet and J.Clot
Laboratoire d'Immunologie-INSERM U291, Hôpital Saint-Eloi, Montpellier, France

Source/Description: Using the published sequence (1), upstream (5-'TGAGGTGTGTTCATTAGTCAACTC-3', −744 to −721) and downstream (5'-GAGCTCGGGAGTGAGGCAGAACA-G-3', +8 to +31) primers were selected for the PCR amplification of human genomic DNA.

Protocol: Reactions were performed using 1 μg of genomic DNA and 50 pmoles of each primer. DNA was amplified for 30 cycles: denaturation, 94°C, 2 min; annealing, 55°C, 2 min; and extension, 72°C, 2 min. Five μl of the reaction was analysed directly on a 1.5% agarose gel run in Tris-borate buffer.

Polymorphism: DdeI digestion of PCR-ampified fragment (773 bp), corresponding to the HLA-DRA regulatory region identified two allelic patterns:
Pattern of allele A1: 482, 158, 69 and 64 bp.
Pattern of allele A2: 274, 208, 168, 69 and 64 bp.

Frequency: Studied in 55 unrelated Caucasian individuals.
Allele A1: 0.73
Allele A2: 0.27

Not Polymorphic For: MaeIII, BstNI, HaeIII, AvaII, BglII, AvaI, HindIII, PstI, SacI, SmaI, TaqI, XhoI.

Chromosomal Localisation: Human MHC, 6p21.

Mendelian Inheritance: Codominant segregation of the DdeI RFLP was observed in four families.

Other Comments: This DdeI polymorphism has not been previously described, and furthermore it represented the first DRA regulatory region polymorphism. This polymorphism in the regulatory region of HLA-DRA could explain the different level of HLA class II cell-surface expression.

Reference: 1) Das,H.K., Lawrance,S.K. and Weismann,S.M. (1983) *Proc. Natl. Acad. Sci. USA* **80**, 3543−3547.

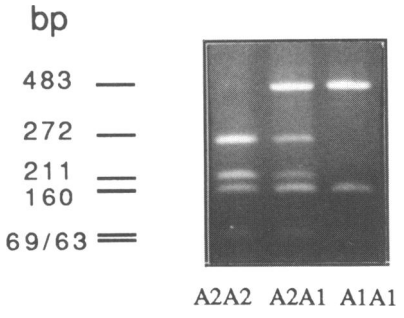

Figure 1. DdeI digestion of HLA-DRA regulatory region amplified by PCR. Examples of the three genotypes are indicated.

A SacI RFLP of the human T-cell receptor delta (TCRD) joining segment J2

D.Alexandre, F.Bernard, C.Buresi, G.Lefranc and M.-P.Lefranc
Université Montpellier II-Laboratoire d'Immunogénétique Moleculaire, URA CNRS 1191, Place Eugene Bataillon, 34095 Montpellier Cedex 5, France

Source/Description: TCRDJ2 probe, R21XH (1), a 2.8 kb XhoI-HindIII fragment containing the TCRDJ2 segment, subcloned in pUC.

Polymorphism: SacI (GAGCT/C) identifies three allelic restriction fragments. One of them (12.0 kb) corresponds to the *TCRDJ2*B1* allele. The other two (3.0 + 9.0 kb) correspond to the *TCRDJ2*B2* allele and result of the presence of a polymorphic SacI site in the region hybridizing to the R21XH probe.

Frequency: *TCRDJ2*B1*: 0.26
**B2*: 0.74
Studied in 35 unrelated Tunisians.

Not Polymorphic For: EcoRI (4.5 + 5.1 kb), HindIII (3.1 kb) (studied in 8 unrelated individuals) (2) and XbaI (4.9 kb) (studied in 26 unrelated Tunisians).

Chromosomal Localization: On chromosome 14 at band 14q11.

Mendelian Inheritance: Co-dominant segregation demonstrated in one family.

Probe Availability: Dr T.H.Rabbitts.

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References: 1) Baer,R., Boehm,T., Yssel,H., Spits,H. and Rabbitts,T.H. (1988) *Embo J.* **7**, 1661−1668. 2) Chuchana,P., Soua,Z., Ghanem,N., Brockly,F., Lefranc,G. and Lefranc,M.-P. (1989) *Nucl. Acids Res.* **17**, 1275.