An MspI polymorphism at the MX1 locus in 21q22.3

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Source and Description: The probe (pBSM132−8b) is a 1.23-kb PstI cDNA fragment in plasmid Bluescript M13 (1). This cloned probe is from the 5′ region of the human MX1 gene which encodes the interferon-induced p78 protein (2). Two PstI RFLPs for the MX1 gene have been described (3).

Polymorphism: The MX1 probe shows a two-allele DNA polymorphism after MspI digestion and Southern blotting with variable DNA fragments A1: 6.0 kb and A2: 7.0 kb.

Frequency: The frequencies of the two alleles were determined in 77 unrelated Caucasians. The frequency of the 6.0-kb allele was 58% and the 7.0-kb allele was 42%. The observed heterozygosity was 0.48.

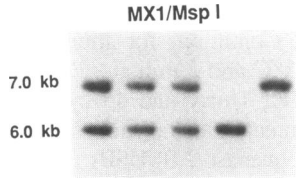
Chromosomal Location: The human MX1 gene maps to 21q22.3 (4).

Mendelian Inheritance: Co-dominant segregation of the restriction fragment length polymorphism was demonstrated in three families (9 individuals).

Probe Availability: Contact Dr. M.A.Horisberger.

Other Comments: We routinely perform genomic MspI digestions at room temperature with 4 units/μg DNA. This polymorphism is observed under normal hybridization and wash conditions.

References: 1) Horisberger,M.A. *et al.* (1990) *J. Virol.* **64**, 1171−1181. 2) Horisberger,M.A. *et al.* (1988) *Somat. Cell Molec. Genet.* **14**, 123−131. 3) Petersen,M.B. *et al.* (1989) *Nucl. Acids Res.* **17**, 7546. 4) Gardiner,K. *et al.* (1990) *EMBO J.* **9**, 25−34.



*Hae*III polymorphism within 3′ untranslated region of PRAD1

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Source/Description: PCR primers for the amplification of a 1223 bp fragment of the 3′ untranslated region of the *bcl*I-linked gene PRAD1 (1).

Primers:
5′ GGAAAGCTTCATTCTCCTTGTTG 3′
5′ TCTAGGTAAACCTCTGAGGTCC 3′

Polymorphism: *Hae*III identifies a simple two allele polymorphism. Constant bands 217, 184 and 130 bp. F1 692 bp; F2 581 and 111 bp.

Frequency: Studied in 44 unrelated individuals
F1 0.35
F2 0.75

Not Polymorphic For: *Rsa*I, *Hinf*I, *Msp*I, *Sau*3a, *Mva*I, *Cfo*I, *Dde*I or *Taq*I in a panel of nine individuals.

Chromosomal Localisation: PRAD1 has been localised to 11q13 within a cluster of genes often amplified in breast carcinomas (1).

Mendelian Inheritance: Co-dominant segregation demonstrated in four families (19 individuals).

PCR Conditions: 1 μg of genomic DNA was amplified in a 100 μl reaction mix containing dNTPs (0.25 mM), 1 μg of each oligo and *Taq* buffer (BCL). After heating for 4 min at 97°C, 2.5 units of *Taq* polymerase (BCL) was added and reactions cycled 30 times at 94°C for 1 min, 61°C for 1 min and 74°C for 3 min.

Comment: As the polymorphism is within a transcribed region of the gene (3′ untranslated) it should allow the PCR based determination of allele specific expression within RNA samples, in addition to the detection of allele imbalance (caused by allele loss or amplification) within a DNA sample.

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Reference: 1) Motokura *et al.* (1991) *Nature* **350**, 512−515.

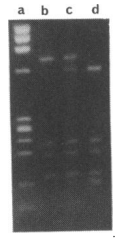

Figure shows amplification products from three individuals digested with *Hae*III. Marker track (a) is a øX174 *Hae*III digest.