Two PstI DNA polymorphisms adjacent to the human gene for the interferon-induced p78 protein (MX1 gene)

Michael B. Petersen¹, Michel A. Horisberger³, Andrew C. Warren^{1,2} and Stylianos E. Antonarakis¹

Genetics Units, ¹Department of Pediatrics and ²Psychiatry, The Johns Hopkins University School of Medicine, Baltimore, MD 21205, USA and ³Ciba-Geigy Ltd, Biotechnology Department, CH-4002, Basel, Switzerland

Source and description of Clone: A 1230 bp PstI cDNA insert in plasmid Bluescript m13⁺ was used as a hybridization probe. This subclone contains upstream non-coding and N-terminal coding sequences of clone p78/2-8b which is the cDNA for the MX1 gene and which encodes an interferon-induced human protein (p78), the equivalent of the interferon-induced murine Mx protein.

Polymorphism: After PstI digestion and Southern blotting the p78/2-8b probe reveals two different DNA polymorphisms. In the first the allelic DNA fragments are A₁: 7.2 kb and A₂: 9.0 kb. A constant fragment of 4.5 kb is also observed. The second polymorphism has allelic DNA fragments of B₁: 1.6 kb and B₂: 1.7 kb. **Frequency:** The frequency of the alleles of the PstI polymorphisms in the CEPH families are as follows:

	n		PIC
PstI A Allele A1	141	0.55	0.372
A2	115	0.45	
PstI B Allele B1	196	0.76	0.295
B2	60	0.24	

Mendelian inheritance: Demonstrated in 40 CEPH families.

Haplotypes: The frequency of the haplotypes for the PstI alleles in the CEPH families are as follows:

	n		PIC
A1 B1	115	0.45	
A1 B2	26	0.10	
A2 B1	81	0.32	0.613
A2 B2	34	0.13	

Linkage disequilibrium: The Δ value between the two polymorphic systems is 0.1306. Therefore there is no significant linkage disequilibrium between them.

Linkage analysis has also been performed using the data from the CEPH families:

$$\begin{aligned} \text{PstI A} - \text{PstI B} \quad \hat{\theta}: 0.011 \quad \hat{z}: 19.57 \\ \hat{\theta}_m: 0.00 \quad \hat{\theta}_f: 0.029 \quad \hat{z}: 19.95 \end{aligned}$$

(There is one definite recombinant in a female meiosis)

Chromosomal localization: The MX1 gene maps on 21q22

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

Reference: Horisberger, MA, et al (1988) Somatic Cell & Mol. Genet. 14:123-131.

Acknowledgements: S.E.A. is supported by NIH grant R01 HD19591. M.B.P. is supported by Danish Research Council & Academy & with Fulbright Fellowship.