TaqI RFLP in the interferon gamma receptor 1 gene (IFNGR1) on human chromosome 6q

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Source/Description: The probe used for Southern hybridization is a 1.8kb IFNGR1 cDNA fragment cloned into the *Sma*I site of pUC19 (pUChGIFR16'; ref.1).

Polymorphism: *Taq*I identifies a two allele RFLP with bands at 6.1kb (A1) and 3.2kb + 2.9kb (A2). Invariant bands are 3.8kb and 1.2kb.

Frequency: Calculated from 33 unrelated Caucasians
A1: 0.85
A2: 0.15
Heterozygosity: 0.18

Not Polymorphic For: *Bgl*II, *Bam*HI, *Eco*RI, *Hind*III, *Msp*I, *Pst*I, *Pvu*II, *Sac*I, *Stu*I, *Xba*I, and *Xmn*I (tested on a panel of 9 unrelated individuals).

Chromosomal Localization: IFNGR1 has been assigned to human chromosome 6q23-q24 by *in situ* chromosomal hybridization (2).

Mendelian Inheritance: Co-dominant segregation demonstrated in 2 informative families.

Probe Availability: American Type Culture Collection.

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References: 1) Aguet,M. *et al.* (1988) *Cell* **55**, 273–280. 2) Le Coniat,M. *et al.* (1989) *Human Genetics* **84**, 92–94.

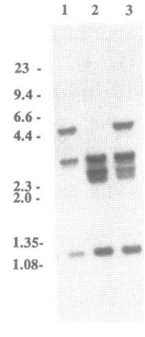

Figure 1. IFNGR1 RFLP detected by Southern blot analysis of TaqI digested human DNA. The two alleles are identified by 6.1kb (A1) and 3.2+2.9kb (A2) bands in a .8% agarose gel. Lane 1: A1 homozygous; lane 2: A2 homozygous; lane 3: heterozygous. Hybridization and washing conditions were 50% formamide, 3× SSC, 37°C and .2× SSC, 60°C, respectively.

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*Pvu*II RFLP in the cytovillin gene (VIL2) on human chromosome 6q

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Source/Description: The probe used for Southern hybridization (pCV6) is a 2.2kb VIL2 cDNA fragment cloned into the *Eco* RI site of pGEM (1).

Polymorphism: *Pvu*II identifies a two allele RFLP with bands at 6.1kb (A1) and 4.8kb (A2). Invariant bands are 8.1kb, 2.1kb, 1.5kb, 1.3kb, and 0.9kb.

Frequency: Calculated from 33 unrelated Causasians:
A1: 0.56
A2: 0.44
Heterozygosity: 0.64

Not Polymorphic For: *Bam*HI, *Eco*RI, *Hind*III, *Hinf*I, *Msp*I, *Pst*I, *Rsa*I, *Sac*I, *Stu*I, *Taq*I, *Xba*I, and *Xmn*I (tested on a panel of 9 unrelated individuals).

Chromosomal Localization: VIL2 has been assigned to human chromosome 6q22-q27 using somatic cell hybrids (1).

Mendelian Inheritance: Co-dominant segregation demonstrated in 3 informative families.

Probe Availability: Contact Dr. Ossi Turunen, Department of Virology, University of Helsinki, SF-00290 Helsinki, Finland

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Reference: 1) Turunen,O. *et al.* (1989) *J. Biol. Chem.* **264**, 16727–16732.

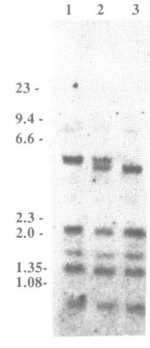

Figure 1. VIL 2 RFLP detected by Southern Blot analysis of *Pvu*II digested human DNA. The two alleles are identified as 6.1kb (A1) and 4.8kb (A2) bands in a .8% ararose gel. Lane 1: A1 homozygous; lane 2: heterozygous; lane 3: A2 homozygous. Hybridization and washing conditions were 50% formamide, 3× SSC, 37°C and .2× SSC, 60°C, respectively.

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