SacI RFLP in the insulin-like growth factor 2 receptor gene (IGF2R) on human chromosome 6q

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Source/Description: The probe used for Southern hybridization is a 4.6 kb *KpnI* fragment derived from the 8.6 kb IGF2R cDNA clone (pSVLhMPR5) (1).

Polymorphism: *SacI* identifies a two allele RFLP with bands at 18.6 kb (A1) and 14.4 kb (A2). Invariant bands are 5.3 kb, 4.9 kb, 4.0 kb, 3.1 kb and 2.2 kb.

Frequency: Calculated from 31 unrelated Caucasians

A1: 0.65

A2: 0.35

Observed heterozygosity: 0.52.

Not Polymorphic For: *AluI*, *BamHI*, *EcoRI*, *HaeIII*, *HindIII*, *HinfI*, *MspI*, *PstI*, *PvuII*, *StuI*, *TaqI* and *XmnI* (tested on a panel of 9 unrelated individuals).

Chromosomal Localization: IGF2R has been assigned to human chromosome 6q25–27 using somatic cell hybrids and *in situ* chromosomal hybridization (2).

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

Probe Availability: Contact Dr William S. Sly, Department of Biochemistry and Molecular Biology, St Louis University School of Medicine.

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References: 1) Oshima, A. *et al.* (1988) *J. Biol. Chem.* **263**, 2553–2562. 2) Laureys, G. *et al.* (1988) *Genomics* **3**, 224–229.

Figure 1. IGF2R RFLP detected by Southern blot analysis of *SacI* digested human DNA. The two alleles are identified as 18.6 kb (A1) and 14.4 kb (A2) bands in a .8% agarose gel. Lane 1: A1 homozygous; lane 2: heterozygous; lane 3: A2 homozygous. Hybridization and washing conditions were 50% formamide, $3\times$ SSC, 37°C and $.2\times$ SSC, 60°C, respectively.

A *PvuII* RFLP at D6S114E locus in the HLA region (RING4 locus)

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Source and Description of Clone: The probe P21U is a 2.8 kb cDNA RING4 clone derived from the U937 cDNA library and subcloned in CDM8 (1).

Polymorphism: *PvuII* identifies a two allele polymorphism with fragments of either 6.8 kb (A1) or 3.9 and 2.9 kb (A2) in length. No other bands are detected.

Frequency: As determined from 76 chromosomes of unrelated subjects of Sardinian origin.

Allele A1 = 0.68

Allele A2 = 0.32

Not Polymorphic For: 9 enzymes including *BamHI*, *BglII*, *EcoRI*, *HindIII*, *MspI*, *PstI*, *RsaI*, *TaqI* and *XbaI*.

Chromosomal Location: The human RING4 gene has been localised to the short arm of chromosome 6, between HLA DOB and DNA genes, about 25 kb centromeric to the former.

Mendelian Inheritance: Mendelian inheritance was demonstrated in three two-generation families tested.

Other Comments: No linkage disequilibrium between the two RING4 alleles and any of the 21 DPB1 or the 9 DQA1 HLA genes was found. Moreover no linkage disequilibrium between any of the two peculiar HLA extended haplotypes present in Sardinia at a very high frequency (2, 3), namely A30, B18, Cw5, DR3, DRw52, DQw2 and A2, Cw7, B17, 3F31, DR2, DQw1, and the two RING4 alleles was observed.

Probe Availability: Contact Dr John Trowsdale, Imperial Cancer Research Fund, Lincoln's Inn Fields, London.

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