Dinucleotide repeat polymorphism at the human GLUT2 locus

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Source and Description: A (dC-dA)n . (dG-dT)n sequence was found within an intron of the human liver/islet glucose transporter (GLUT2) gene. Two oligonucleotides homologous to the sequences flanking the dinucleotide repeat were used to amplify the region and generate a fragment of the predicted size of 112 bp.

Primer Sequences:
5′ CTCCAAGAAGCATATCAGGA 3′ (CA strand)
5′GTCCACATACCGCCTTTAGAG 3′(GT strand)

Frequencies: Estimated from 44 chromosomes of unrelated Caucasian family parents.

Allele (bp)	Frequency	Allele (bp)	Frequency
K1 124	0.05	K3 120	0.54
K2 122	0.23	K4 116	0.18

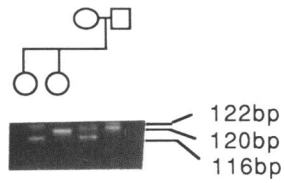
Chromosomal Localization: Assigned to chromosome 3q26.1−3q26.3 using a panel of mouse-human somatic cell hybrids and by in situ hybridisation to metaphase chromosomes (1).

Mendelian Inheritance: Co-dominant segregation observed in two Caucasian families (38 individuals).

Other Comments: The PCR reaction was performed on 1 μg genomic DNA as previously described (2) except that samples were processed through 30 temperature cycles consisting of 2 min at 94°C, 2 min at 55°C and 3 min at 72°C. PCR products were fractionated on 10% PAGE gel and visualized by staining with ethidium bromide. The sizes of the alleles were determined by comparision with *MspI* digested *pBR*322 DNA. The sequence of GLUT2 can be obtained from Dr.G.I. Bell.

Acknowledgements: This work was supported by a grant from the Medical Research Council (U.K.).

References: 1) Fukumoto,H. *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 5434−5438. 2) Saiki,R.K. *et al.* (1988) *Science* **230**, 487−491.



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*Sma*I and *Hha*I polymorphisms in the 5′ region of the human von Willebrand factor gene (F8VWF)

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Description: Comparison of genomic DNA sequences (1) predicted three polymorphisms, *Sma*I, *Hha*I and *Bsp*HI within a 1.46 kb amplified segment of the human vWF gene spanning exon 3.

Primer Sequences:
5′ AGG TAG TTT GCA CAA GTT GGT CAC 3′
5′ CAA CCT TCA TGA GCC TTG GTT CTC 3′

Methods: PCR conditions: 10 mM Tris-HCl pH 8.3, 50 mM KCl, 1.5 mM $MgCl_2$, 0.5 μM primers, 200 μM dNTP, 0.5 μg DNA, 10% DMSO and 1 U *Taq* polymerase to 100 μl. 36 cycles: 94°C (1 min), 65°C (2 mins) and 72°C (1.5 mins).

*Sma*I *Polymorphism*: Position 4996 (1).
Allele Q1: 1109 bp; Q2: 857, 252 bp. Constant band; 352 bp.

*Hha*I *Polymorphism*: Position 5195 (1).
Allele R1: 1156 bp; R2: 1010, 146 bp. Constant band; 305 bp.

*Bsp*HI Polymorphism: Position 5971 (1).
No *Bsp*HI site detected in 80 chromosomes analysed.

Allele Frequencies: (176 chromosomes examined).
Q1 and R1 show complete linkage disequilibrium; frequencies at both *Sma*I and *Hha*I sites are allele 1: 0.34; allele 2: 0.66.
Polymorphism Information Content = 0.35
In heterozygotes double digestion confirmed that Q1 and R1 lay on the same chromosome. No Q1,R2 or Q2,R1 haplotypes were observed.

Mendelian Inheritance: Co-dominant segregation was demonstrated in 26 meioses in 2 families.

Chromosomal Localization: 12p12−12pter (2).

Acknowledgements: This research was supported by the Ludovici Bequest Fund to the University of Edinburgh.

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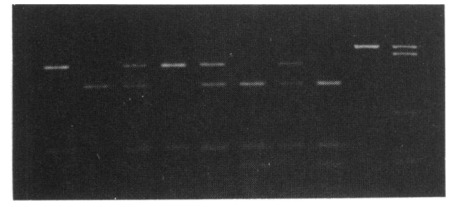

Figure 1. 1.4% agarose gel showing *Sma*I digestion products (undigested product — lane 9). Marker (10) is *Taq*I digested pBR322.