BamHI RFLP at the GLUT3 locus

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Source and Description: pBS/MGT3 is a cDNA clone containing a 2.6 kb SalI insert which encodes human fetal skeletal glucose transporter-like protein.

Polymorphism: BamHI identifies a two allele polymorphism with fragments of 5.5 kb (allele A) and 5.0 kb (allele B) and detects four invariant bands of 24 kb, 23 kb, 14 kb and 7 kb.

Frequency: Studied in 16 normal unrelated European Caucasian individuals.
Allele A = 0.6 kb
Allele B = 0.4 kb

Chromosomal Localization: Assigned to chromosome 12p13.3 using a panel of mouse-human somatic cell hybrids and by in situ hybridization to metaphase chromosomes (1).

Mendelian Inheritance: Co-dominant segregation observed in 2 families.

Probe Availability: Contact G.I.Bell.

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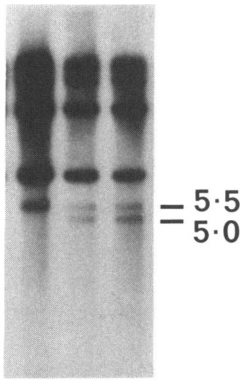
Reference: 1) Kayano,T. *et al.*(1988) *J. Biol. Chem.* **263**, 15245–15248.



TG repeat polymorphism at the D21S167 locus

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Source and Description: Cosmid ICRFc102A0322 was from a flow-sorted human chromosome 21-specific library. DNA sequences flanking a $(GT)_{19}$ repeat within this cosmid (EMBL accession number X52289) were used to design PCR primers.

PCR Primers:
(#112)-5′-TCCTTCCATGTACTCTGCA-3′
(#113)-5′-TGCCCTGAAGCACATGTGT-3′

Polymorphism: Allelic fragments were resolved on DNA sequencing gels. Lengths of allelic fragments (nt) were: A1 = 156, A2 = 158, A3 = 160, A4 = 162, A5 = 164, A6 = 166, A8 = 172, A9 = 174, A10 = 176, A11 = 178, A12 = 180, A13 = 182.

Frequencies: Allele frequencies in 33 unrelated European Caucasians: A1 = .030, A2 = .272, A3 = 288, A4 = .061, A5 = .030, A6 = .030, A7 = .061, A8 = .061, A9 = .030, A10 = .061, A11 = .045, A12 = .015, A13 = .015; PIC = 0.80.

Chromosomal Localization and Mendelian Inheritance: PCR of genomic DNAs from a somatic cell hybrid panel (1) indicated localization to 21q22.2. Linkage analysis in 3 CEPH families showed no recombination in 21 informative phase-known meioses with D21S15 (previously localized to 21q22.2 (1)), giving a maximum LOD score of 6.3 at theta = 0. Mendelian inheritance was observed in all cases.

PCR Conditions: We carry out PCR as in reference (2) using an annealing temperature of 56°C.

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References: 1) Gardiner,K., Horisberger,M., Kraus,J., Tantravahi,U., Korenberg,J., Rao,V., Reddy,S. and Patterson,D. (1990) *EMBO J.* **9**, 25–34. 2) Luty,J.A., Guo,Z., Willard,H.F., Ledbetter,D.H., Ledbetter,S. and Litt,M. (1990) *Am. J. Hum. Genet.* **46**, 776–783.

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