Four RFLPs at the D8S42 locus

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Source/Description: L1 is a 125 bp single copy genomic DNA fragment cloned into pUC13. The clone was isolated by microdissection and microcloning of the Langer-Giedion syndrome chromosome region (LGCR).

Polymorphisms: L1 identifies four two-allele polymorphisms:

HindIII	9.2 kb A1	EcoRV	21.0 kb B1
	4.4 kb A2		23.0 kb B2
TaqI	1.3 kb C1	PstI	18.0 kb D1
	2.1 kb C2		5.4 kb D2

Frequencies:

Allele	Frequency	Number of Chromosomes
A1	0.81	32
A2	0.19	
B1	0.72	40
B2	0.28	
C1	0.81	36
C2	0.19	
D1	0.81	32
D2	0.19	

Not Polymorphic For: AccI, ApaI, AvaI, AvaII, Asp700, BanI, BanII, BamHI, BclI, BglI, BglII, BstNI, BstXI, DraI, EcoRI, HindII, HinfI, HpaI, KpnI, MspI, NciI, PvuII, RsaI, SacI, Sau96I, StuI, XbaI in a screen of six unrelated Caucasian individuals.

Chromosomal Localization: Mapped to 8q23.2-q24.11 by quantitative Southern blot hybridization of DNA from patients with Langer-Giedion syndrome that have cytogenetically visible deletions.

Mendelian Inheritance: Autosomal codominant segregation was observed in two large Caucasian families.

Probe Availability: Available for collaboration: contact B.Horsthemke.

Other Comments: Insert can be released from the vector by EcoRI digestion.

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Reference: Lüdecke *et al.* (1989) *Nature* **338**, 348–350.

Two EcoRI RFLPs at the GLUT2 locus

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Source/Description: A full length human cDNA clone for the liver type glucose transporter, pHTL3/BS (kindly provided by G.Bell).

Polymorphism: EcoRI identifies two 2-allele restriction fragment length polymorphisms with fragment sizes 2.5 kb (A1) or 2.3 kb (A2), and 4.2 kb (B1) or 4.0 kb (B2). Constant bands of 9.2, 7.2 and 5.6 kb are also seen.

Frequency: Estimated from 52 unrelated Scandinavians.

A1 (2.5 kb): 0.11

A2 (2.3 kb): 0.89

B1 (4.2 kb): 0.81

B2 (4.0 kb): 0.19

Other Enzymes Tested: The 2.5/2.3 kb polymorphism was also detected with PstI, SacI, HindIII and TaqI.

Mendelian Inheritance: Codominant inheritance was shown in 19 informative meioses.

Chromosomal Localization: The q26.1-q26.3 region of chromosome 3 (1).

Probe Availability: Contact Graeme I.Bell, Howard Hughes Medical Institute, University of Chicago, or Holger Luthman.

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Reference: 1) Fukumoto, H. *et al.* (1989) *Proc. Natl. Acad. Sci. USA* **85**, 5434–5438.

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