MspI and SstI RFLPs at the human insulin receptor locus on chromosome 19

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Source/Description: pINSR HS0.7 is a 700 bp Hind III to Sac I genomic fragment of the human insulin receptor gene which includes all of exon 17, 370 bp of 5' intron, and 78 bp of 3' intron. The fragment is cloned in pUC 19. It has been completely sequenced and a map of the region described (Elbein, 1989).

Polymorphism: MspI detects an invariant band at 0.8 kb and bands at 0.6 kb (allele 1) or 1.7 kb (allele 2). SstI detects a band at 9.6 kb (allele 1) or 7.0 kb (allele 2).

Frequency: Studied in 45 unrelated North American Caucasians of predominantly Northern European descent.

MspI	SstI
Allele 1 (0.6 kb) 0.44	Allele 1 (9.6 kb) 0.08
Allele 2 (1.7 kb) 0.56	Allele 2 (7.0 kb) 0.92

Not Polymorphic For: EcoRI, PvuII, RsaI, TaqI, XbaI.

Chromosomal Location: INSR; 19p 13.2–13.3.

Mendelian Inheritance: Segregation was confirmed with previously reported INSR RFLPs in 5 large pedigrees, 2 of which had 3 generations.

Probe Availability: Write to S.C.Elbein.

Other Comments: MspI RFLP is useful in many cases where other RFLPs fail to distinguish alleles. The probe is of unique sequence and these RFLPs present no difficulties. Preliminary data suggest low levels of linkage disequilibrium with previously defined RFLPs. The SstI RFLP appears to represent the same RFLP described earlier with the cDNA probe 12.1P1.6 (S2; Elbein *et al.*, 1986). Sequence and map data place the SstI RFLP 7 kb downstream from the 5' end of this probe, and the MspI RFLP 0.35 kb downstream from exon 17. Mapping data suggest that this probe should detect the previously described Bgl2 RFLP and the 0.5 kb insertion approximately 5.5 kb upstream from this probe with HindIII.

References: Elbein, S.C. *et al.* (1986) *Proc. Natl. Acad. Sci. USA* **83**, 5223, Elbein, S.C. (1989) *Diabetes* **38**, 737.

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