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Four RFLPs of the human insulin receptor gene: PstI, KpnI, RsaI (2 RFLPs)

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SOURCE/DESCRIPTION: Fragments were isolated from subclones containing the insulin receptor cDNA described by Ullrich et al., 1985. Probe 1 as obtained from an SP64 subclone containing the 1011bp EcoRI fragment from the 5' region of the Ullrich cDNA. Probe 1 was a 677bp XhoI/EcoRI fragment from the α subunit region of the IR cDNA corresponding to nucleotides 334 to 1011, the putative ligand binding domain. Probe 2 was obtained from an SP64 subclone containing the 4190bp EcoRI fragment from the 3' end of the Ullrich cDNA. Probe 2 was a 1599bp PstI fragment from the β subunit region of the insulin receptor cDNA corresponding to nucleotides 2746 to 4345, encoding the tyrosine kinase domain.

POLYMORPHISMS: Probe 1: α subunit
PstI identifies a 2 allele polymrophism with bands at 15.2 or 13.7Kb.
RsaI identifies a 2 allele polymorphism with bands at 0.9 or 0.3Kb.
Probe 2: β subunit
KpnI identifies a 2 allele polymorphism with bands at 15.9 or 11.2 and 4.7Kb.
RsaI identifies a 2 allele polymorphism with bands at 1.9 or 0.4Kb.

FREQUENCY: Probe 1: α subunit

PstI	78 Caucasians:	15.2Kb allele, 0.88; 13.7Kb allele, 0.12.
	32 Blacks:	15.2Kb allele, 0.87; 13.7Kb allele, 0.13.
RsaI	16 Caucasians:	0.9Kb allele, 0.87; 0.3Kb allele, 0.13.

Probe 2: β subunit

KpnI	60 Caucasians:	11.2, 4.7Kb allele, 0.68; 15.9Kb allele, 0.32.
RsaI	20 Caucasians:	1.9KB allele, 0.55; 0.4Kb allele, 0.45.
	18 Blacks:	1.9Kb allele, 0.56; 0.4Kb allele, 0.44.

NOT POLYMORPHIC FOR: Probe 1: BclII, KpnI, BclI, PvuII, ScaI in at least 30 chromosomes. Probe 2: PstI, PvuII in at lest 30 chromosomes.

CHROMOSOMAL LOCALISATION: 19p 13.3-19p 13.2 (Yang Feng et al., 1985).

PROBE AVAILABILITY: Write to R. Taub.

MENDELIAN INHERITANCE: Segregation in at least one family was observed for the PstI, KpnI, and RsaI (β) polymorphisms.

REFERENCES:
Ullrich A. et al., Nature (1985) 313, 756-761.
Yang Feng et al., Science (1985) 228, 728-731.

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