Human insulin receptor RFLPs detected by HindIII and DraI

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SOURCE/DESCRIPTION: Two subfragments isolated from the 4.2 kb insulin receptor cDNA clone phINSR 13-1, (Shaw et al., 1985), were used. Probe 13-14 is a 1360 bp BglI/BglI fragment corresponding to nucleotides 1606 to 2965 of the α and β chain precursor cDNA. Probe 13-16 is a 1599 bp PstI/PstI subfragment corresponding to nucleotides 2747 to 4345, encoding the tyrosine kinase domain of the β subunit.

POLYMORPHISMS: Two HindIII RFLPs are detected with the probe 13-14, one two-allele polymorphism with fragment sizes 11.2 kb and 10.4 kb, and one two-allele polymorphism with fragment sizes 14.0 kb and 13.5 kb. The latter polymorphism is also revealed by probe 13-16, together with a third two-allele RFLP with the allele sizes 23 kb and 20 kb.

Two DraI RFLPs are detected with the probe 13-16, one two-allele polymorphism with fragment sizes 2.9 kb and 1.4 kb, and one two-allele polymorphism with fragment sizes 4.1 kb and 3.6 kb.

FREQUENCY: Ascertained in unrelated Scandinavians.

HindIII:	(N= 33)	11.2 kb allele: 0.61	10.4 kb allele: 0.39
	(N= 33)	14.0 kb allele: 0.18	13.5 kb allele: 0.82
	(N= 43)	23 kb allele: 0.15	20 kb allele: 0.85
DraI:	(N= 64)	2.9 kb allele: 0.38	1.4 kb allele: 0.62
	(N= 68)	4.1 kb allele: 0.20	3.6 kb allele: 0.80

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

MENDELIAN INHERITANCE: Co-segregation with insulin receptor haplotypes defined by earlier published insulin receptor RFLPs (HMG 9) was observed in at least four two-generation families (20 individuals).

PROBE AVAILABILITY: Write to H. Luthman or G.I. Bell.

OTHER COMMENTS: In 28 tested unrelated individuals, the 14.0 kb and 13.5 kb HindIII polymorphism and the 4.1 kb and 3.6 kb DraI polymorphism are identical to the earlier described 5.8 kb and 5.3 kb SacI polymorphism.

REFERENCES:

- Shaw D.J. et al. (1985) Nucleic Acids Res. **13**, 8661.
Yang-Feng T.L. et al. (1985) Science **228**, 728-731.

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