DraI and PstI RFLPs in the tyrosine hydroxylase (TH) and insulin gene (INS) region of chromosome 11

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SOURCE/DESCRIPTION: Probes were isolated from the 6.0 kb human genomic clone phINS 500, extending from 7 kb to 13 kb upstream of the insulin gene. Cleavage of the phINS 500 insert with DraI generates two fragments; a 3.2 kb fragment, 500-32, distal to the insulin gene, and a proximal 2.8 kb fragment, 500-28. The probe 500-5 is a 0.5 kb PstI/PstI subfragment of phINS 500.

POLYMORPHISMS: With the probe 500-32 DraI detects a two-allele polymorphism with fragment sizes 10.3 kb (allele 1) and 8.7 kb and 1.6 kb (allele 2). The two longer DraI fragments (10.3 kb and 8.7 kb) are also identified with the probe 500-5. In addition, the probe 500-5 reveals a three-allele PstI polymorphism with fragment sizes 2.8 kb (allele 1), 1.5 kb (allele 2) and 0.5 kb (allele 3).

FREQUENCY: Ascertained in unrelated Scandinavians.

DraI: (N=168) allele 1: 0.63 allele 2: 0.37

PstI: (N=110) allele 1: 0.08 allele 2: 0.29 allele 3: 0.63

CHROMOSOMAL LOCALISATION: 11p15.5 (HGM 9).

MENDELIAN INHERITANCE: Co-dominant segregation was observed for both RFLPs in at least ten two-generation families (42 individuals).

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

OTHER COMMENTS: With the probe 500-28 another DraI RFLP was detected with allele sizes 12.0 kb and 10.5 kb. This DraI RFLP is identical to the VNTR polymorphism located 5' of the insulin gene (Bell et al., 1981). The PstI RFLP described here is different from the two-allele PstI RFLP detected by the tyrosine hydroxylase probe pHGTH4 (Kelsoe et al., 1988).

REFERENCES:

Bell G.I. et al. (1981) Proc. Natl. Acad. Sci. USA **78**, 5759-5763.

Kelsoe J.R. et al. (1988) Nucleic Acids Res. **16**, 7760.

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