A HincII RFLP in the human D4 dopamine receptor locus (DRD4)

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Source/Description: Probe pB28 (1) containing 1.5 kb coding sequence encoding the dopamine D4 receptor gene (DRD4).

Polymorphism: Digestion with HincII reveals a 4 allele polymorphism with fragment sizes 6.5 kb (A1), 4.6 kb (A2), 4.3 kb (A3) and 4.0 kb (A4). No constant bands are observed.

Frequency: Estimated from 36 unrelated Caucasians of Northern European descent.
A1: 0.04
A2: 0.15
A3: 0.20
A4: 0.61
The observed heterozygosity was 0.44.

Chromosomal Location: Determined from genetic linkage analysis to be near 11pter; lod scores >11 with HRAS (2).

Mendelian Inheritance: Co-dominant inheritance was observed in 7 informative Caucasian families.

Availability: Contact Dr H.H.M. Van Tol, Departments of Pharmacology and Psychiatry, University of Toronto, Toronto, Ontario, M5S 1A8, Canada.

Other Comments: There is a repeat sequence in the probe necessitating addition of total human DNA to the hybridization solution.

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References: 1) Van Tol,H.H.M. *et al.* (1991) *Nature* **350**, 610–614. 2) Gelernter *et al.* (1991) *Amer. Soc. Hum. Genet.* (Abstract in press).

*Bcl*I RFLP at the NF1 gene locus

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Source/Description: Cawthon,R.M. *et al.* (1) reported that the NF1 gene is organized into 11 or more exons. The probe we used was a 424 bp fragment amplified by Polymerase Chain Reaction from genomic DNA, which includes the exon 4 of the NF1 gene described in their paper. The primers used were also described in their paper. The forward primer was 5′-ATAATTGTTGATGTGATTTTCATTG-3′ (from nt 58 to 35 upstream of the exon 4), and the reverse primer was 5′-AATTTTGAACCAGATGAAGAG-3′ (from nt 172 to 152 downstream of the exon 4).

Polymorphism: After *Bcl*I digestion of genomic DNA, the probe identified a two-allele polymorphism with bands at (F1) 7.0 kb and (F2) 6.5 kb. No non-polymorphic bands were identified.

Frequency: Estimated from 50 unrelated Japanese individuals.
7.0 kb allele (F1) 0.4
6.5 kb allele (F2) 0.6

Observed Frequency of Heterozygosity: 0.52.

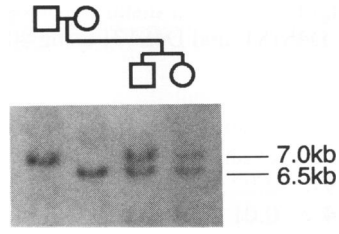
Not Polymorphic For: *Sau*3A-I, *Stu*I, *Rsa*I, *Pst*I, *Xmn*I, *Bam*HI, *Hin*dIII, *Eco*RI and *Hae*III (studied in 25 unrelated Japanese individuals).

Chromosomal Localization: 17q11.2.

Mendelian Inheritance: Co-dominant segregation demonstrated in four 2 generation families (8 meioses).

Clinical Relevance: Prenatal and carrier diagnosis. Analysis for loss of heterozygosity.

Reference: 1) Cawthon,R.M. *et al.* (1990) *Cell* **62**, 193–201.



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