TaqI identifies a four allele DNA polymorphism of the human endothelin-1 gene (EDN1)

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Source/Description: A 1.2 kb EcoRI fragment of the endothelin-1 (EDN1) cDNA, derived from the 5' end, was used as a probe. The EDN1 cDNA consists of this 1.2 kb and at least two identical 0.9 kb EcoRI fragments, inserted in pUC13 (1).

Polymorphism: Hybridization of human genomic DNA digested with TaqI identifies a four allele polymorphism with bands at 5.8 kb (allele 1), 7.7 kb (allele 2), 3.4 and 2.0 kb (allele 3), 3.1 and 2.4 kb (allele 4).

Frequency: Evaluated in 98 unrelated Caucasians: Allele 1: 0.694; Allele 2: 0.265; Allele 3: 0.005; Allele 4: 0.036.

Not Polymorphic For: BglII, DraI, PstI, SacI, Sau96, StyI.

Chromosomal Localization: The EDN1 gene has been assigned by somatic cell hybrid analysis to human chromosome 6 (1). We have obtained additional information on the localization of this gene through genetic mapping evidence for linkage was given to loci F13A and D6S7 (which are located at the tip of chromosome 6) at recombination fractions = 0.07, and to various HLA markers at recombination fractions = 0.30 (3).

Mendelian Inheritance: Codominant segregation has been observed in 27 two- and three-generation families from the CEPH (Centre d'Etude du Polymorphisme Humain) reference panel, and additionally in 13 medium-sized disease families (about 16 members per pedigree).

Other Comments: High stringency wash of the blots in $0.1 \times \text{SSC}/0.1\%$ SDS at 65°C showed no significant background. The digests were run on 0.8% agarose gels on a constant field electrophoresis at 40 V. An additional constant band was observed at 1.8 kb.

A different, two allele Taq I RFLP of this gene at the EDN1 locus was reported by Berge and Berg, 1990 (2).

Probe Availability: The ET1 probe was kindly provided by Dr. Kenneth D.Bloch, Department of Medicine, Harvard Medical School and Cardiac Unit, Massachusetts General Hospital, Boston, MA, USA.

References: 1) K.D.Bloch *et al.* (1989) *J. Biol. Chem.* **264**, 10851–10857. 2) K.E.Berge and K.Berg (1990) *Nucl. Acids Res.* **18**, 6176. 3) M.R.Hoehe and H.Ehrenreich, in preparation.

A TaqI RFLP at the DRD1 locus

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Source and Description of Clone: pHD1-2B is a 1.35 kb Sau3A fragment of the human genomic dopamine D1 receptor clone HGR213-1 (1). The 5' protruding ends of this fragment were half filled in prior to ligation into the half filled in SalI site of pTZ18u.

Polymorphisms: TaqI identifies a 2-allele RFLP with A1=6.6, A2=3.1 kb.

Frequencies: Studied in 65 unrelated CEPH parents. A1=.11, A2=.89.

Not Polymorphic With: ApaI, ApaLI, Asp700, BamHI, BclI, BglII, BstEII, DraI, EcoRI, EcoRV, HindIII, MspI, PstI, PvuII, SacI, ScaI, StuI.

Chromosomal localization: Clone HGR213-1 previously localized to 5q35.1, by fluorescence in situ hybridization (1).

Mendelian Inheritance: Codominant Mendelian inheritance was observed in 4 CEPH families with a total of 34 children.

Availability: Available for collaboration: write to O.Civelli.

Other Comments: The TaqI RFLP is in linkage equilibrium with the previously reported EcoRI RFLP (1). Twenty-one of 65 (32%) of unrelated CEPH parents were heterozygous at one or both of these loci.

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References: 1) Grandy,D.K., Zhou,Q., Allen,L., Litt,R., Magenis,R.E., Civelli,O. and Litt,M. (1990) *Am. J. Hum. Genet.* **47**, 828–834.