Dinucleotide repeat polymorphism in the human thyroid hormone receptor β gene (THRB) on chromosome 3

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Primers/Description: Two primers (THRB-1, 5'-GATCACAA-GGATGCTAGAGT-3', and THRB-2, 5'-TCAAAGGAGTCA-GGCTGTAG-3') were used to amplify a 197–209 bp CA repeat-rich region in intron 2 between Exon 2 (Exon B) and Exon 3 (Exon C) of the human THRB gene (1).

Frequency: Five alleles were observed in 46 unrelated Caucasians. The heterozygosity was 50%.

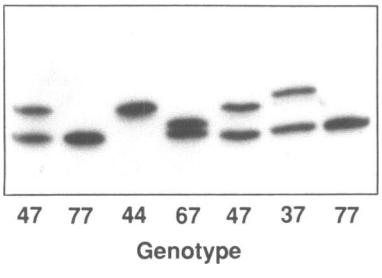
Allele	bp	Frequency
G1	209	0.01
G2	205	0.01
G3	203	0.23
G4	199	0.07
G5	197	0.68

Chromosomal Localization:: THRB has been localized to chromosome 3p24 (2).

Mendelian Inheritance:: Co-dominant inheritance was observed in two families in which 33 individuals were typed.

Other Comments: The PCR was performed using ^{32}P-labeled THRB-1 and unlabeled THRB-2 for 30 cycles; denaturation at 94°C for 1 min; annealing at 55°C for 2 min; and extension at 72°C for 2 min. The PCR products were analyzed on a 5% denaturing polyacrylamide gel (Figure). The amplified dinucleotide repeat sequence was of the form $(CA)_3CGTG(CA)_7T(AC)$; the complete sequence of this region is available from the authors.

References: 1) Sakurai,A. *et al.* (1990) *Mol. Cell. Endocrinol.* **71**, 83. 2) Albertson,D.G. *et al.* (1989) *Hum. Genet.* **83**, 127.



PCR amplification of dinucleotide repeat DNA polymorphism in human THRB. The genotypes are noted at the bottom of the figure.

An EcoRI polymorphism for the PLAUR gene

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Source/Description: puPAR-2 is a 600 bp BamHI cDNA fragment subcloned in the vector pEMBL 18 from the puPAR-1 cDNA clone of the PLAUR gene (1).

Polymorphism: EcoRI identifies a two-allele restriction fragment length polymorphism (RFLP) with allelic bands at 4.2 kb (A1) and 4.4 kb (A2) (fig. 1). There are two constant bands at 9.0 kb and 2.3 kb (not shown in figure).

Frequency: Allele frequencies are based on 63 of the unrelated European CEPH parents (i.e. Venezuelans and related parents excluded):
4.2 kb allele (A1) 0.92
4.4 kb allele (A2) 0.08
10% of the analysed individuals showed heterozygosity.

Not Polymorphic For: ApaI, BamHI, BanII, BclI, BglII, BstEII, DraI, EcoRV, HindIII, KpnI, MspI, PvuII, RsaI, SacI, TaqI, XbaI using a panel of 6 unrelated Caucasians.

Chromosomal Location: In a location analysis using a panel of human/rodent cell hybrids and in a multipoint linkage analysis of 40 CEPH families puPAR-2 was found to be located on chromosome 19q13.1-q13.2 (2).

Mendelian Inheritance: Mendelian inheritance was observed in 10 informative CEPH families, a total of 98 individuals.

Probe Availability: Available for collaboration.

Other Comments: An infrequent and faint 2.6 kb band was variably present (not shown in figure) and probably due to star activity of the enzyme.

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References: 1) Roldan,A.L., Cubellis,M.V., Masucci,M.T., Behrendt,N., Lund,L.R., Danø,K. and Blasi,F. (1990) *EMBO J.* **9**, 467–474. 2) Børglum,A.D., Byskov,A., Ragno,P., Roldan,A.L., Tripputi,P., Cassani,G., Danø,K., Blasi,F., Bolund,L. and Kruse,T.A. (1991) *Am. J. Hum. Genet.* in press.

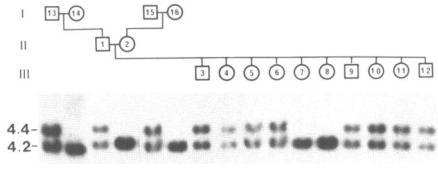

Figure 1. The EcoRI polymorphism in a Southern blot of Centre d'Etude du Polymorphisme Humain family 104.