Trinucleotide repeat polymorphism at the human transcription factor IID gene

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Source/Description: The polymorphic $(CAG)_n$ repeat begins at base pair 226 of the human transcription factor IID gene on chromosome 6 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 203 bp.

Primer Sequences:
GACCCCACAGCCTATTCAGA (CAG strand);
TTGACTGCTGAACGGCTGCA (GTC strand).

Frequency: Estimated from 48 chromosomes of unrelated individuals.
Observed heterozygosity = 81%. PIC = 0.78

Allele (bp)	Frequency	Allele (bp)	Frequency
A1 206	0.02	A5 194	0.17
A2 203	0.08	A6 191	0.15
A3 200	0.27	A7 185	0.08
A4 197	0.23		

Mendelian Inheritance: Co-dominant segregation was observed in two informative families.

Chromosomal Localization: We have tentatively assigned the human transcription factor IID gene to chromosome 6 using rodent/human somatic cell hybrids.

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described except that the denaturation cycle at 94°C was extended to 1.4 minutes. The trinucleotide repeat was based on a $(CAG)_{18}$ sequence.

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*Rsa*I polymorphism in the human δ-aminolevulinate dehydratase gene at 9q34

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Source/Description: Southern hybridization analysis of the *Rsa*I RFLP was carried out with a 1.17 kb human δ-aminolevulinate dehydratase (ALAD; PBG-S; porphobilinogen synthase; EC 4.2.1.24) cDNA clone isolated from an adult liver library (1). For analysis of the *Rsa*I polymorphism by the polymerase chain reaction, oligonucleotide primers were used to amplify a 916 nt region including exon 4 and adjacent intronic sequences.

PCR Primers:
Sense 5′-AGACAGACATTAGCTCAGTA-3′;
Antisense 5′-GGCAAAGACCACGTCCATTC-3′.

Polymorphism: Southern hybridization analysis of *Rsa*I-digested genomic DNA using radiolabeled human ALAD cDNA identified a simple two allele polymorphism located 3.4 kb upstream of the polyadenylation signal. Restriction fragment lengths were 3.0 (A1) or 2.2 (A2). PCR amplification and *Rsa*I digestion yielded a single 916 nt (A1) product or 523 and 393 nt products (A2).

Frequency: Analysis of genomic DNAs from 180 unrelated Caucasian individuals revealed allele frequencies of 0.75 for A1 and 0.25 for A2.

Not Polymorphic For: *Bam*HI, *Bgl*I, *Bgl*II, *Eco*RI, *Hin*dIII, *Nco*I, *Pst*I, *Pvu*II or *Sac*I.

Chromosomal Localization: The ALAD gene has been localized to 9q34 by *in situ* hybridization and by analysis of somatic cell hybrids (2).

Mendelian Inheritance: Co-dominant segregation of the A1 and A2 alleles has been demonstrated in three 3-generation families.

PCR Conditions: Genomic DNA from 0.5 ml blood was amplified for 30 cycles using denaturation at 94°C for 1 min, annealing at 55°C for 1 min and extension at 72°C for 3 min with the final cycle extended to 7 min.

Acknowledgements: We thank Daphne Wong for carrying out PCR analyses. This work was supported by grants DK26824 and ES05046 from the NIH.

References: 1) Wetmur,J.G. *et al.* (1986) *Proc. Natl. Acad. Sci. USA* **83**, 7703–7707. 2) Potluri,V.R. *et al.* (1987) *Human Genet.* **76**, 236–239.

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