Unusual length polymorphism in human steroid 5α-reductase type 2 gene (*SRD5A2*)

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Source/Description: A length polymorphism involving different numbers of dAT dinucleotides and alterations just 5′ to these repeats exists in the last exon (3′-untranslated region) of the *SRD5A2* gene. Mutations in this gene cause male pseudohermaphroditism (1,2) and may also underlie other disorders of androgen metabolism. Amplification of the polymorphic region from genomic DNA with PCR primers h5α2-31 and h5α2-30 followed by gel electrophoresis on 5% (w/v) non-denaturing polyacrylamide gels results in the detection of fragments of different lengths.

PCR Primers: 5′ end nucleotide (1).
h5α2-31: 5′-GCTGATGAAAACTGTCAAGCTGCTGA-3′ 829
h5α2-30: 5′-GCCAGCTGGCAGAACGCCAGGAGAC-3′ 962

875 929

DNA Sequences:
$(TA)_{18}$ allele ... AATAGTCA$(TA)_{18}$ TGTATATATGT ...
$(TA)_9$ allele ... AATAGACC$(TA)_9$ TGTATATATGT ...
$(TA)_0$ allele ... AATAGTCCCGG GTATATATGT ...

Frequency: From analysis of 172 chromosomes: $(TA)_{18}$ allele=0.01; $(TA)_9$ allele=0.02; $(TA)_0$ allele=0.96.

Chromosomal Location: 2p23 (2).

Mendelian Inheritance: Not yet demonstrated due to low frequency of variant alleles.

PCR Conditions: PCR reaction: 10 mM Tris−HCl, pH 8.3; 50 mM KCl; 2 mM $MgCl_2$; 1.75 μmol each primer; 50 μM each dNTP; 3 units of Amplitaq; 0.1 μg genomic DNA; total volume=20 μl; 35 cycles of 94°C 1 min, 68°C 1 min.

Other Comments: Heterozygosity not yet demonstrated, presumably due to low variant allele frequency.

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References: 1) Andersson,D. *et al.* (1991) *Nature* **354**, 159−161. 2) Thigpen,A.E. *et al.* (1992) *J. Clin. Invest.* **90**, 799−809.

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*Taq*I RFLP at norepinephrine transporter protein (NET) locus

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Source/Description: Probe pNET is a 1.9 kb human cDNA in Bluescript SKII (1).

Polymorphism: *Taq*I identifies a three-allele (codominant) polymorphism, with bands at 7.5, 6.8, and 4.6 kb (alleles A1, A2, and A3 respectively). There are also constant bands at 6.2, 5.5, 3.4, 3.3, and 2.2 kb.

Frequency: Among 44 unrelated Caucasians (of Northern European origin): A1 allele (7.5 kb), 0.08; A2 allele (6.8 kb), 0.82; and A3 allele (4.6 kb), 0.10. Observed heterozygosity = 0.36.

Not Polymorphic For: BamHI; BanI; BanII; DraI; EcoRI; HaeIII; HincII; KpnI; PvuII; RsaI; SacI; XbaI; and XmnI, using a panel of 12 unrelated individuals.

Chromosomal Localization: Presently unknown.

Mendelian Inheritance: Codominant inheritance was observed in four extended kindreds.

Probe Availability: Available from S.Amara, Oregon Health Sciences University.

Other Comments: Standard hybridization and wash conditions usually produce sharp RFLP bands with little background.

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Reference: 1) Pacholczyk,T., Blakely,R.D. *et al.* (1991) *Nature* **350**, 350−354.

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