Dinucleotide repeat polymorphism at the D14S43 locus

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Source/Description: Cosmid ICRFc102E1222 was from a flow sorted chromosome 21 cosmid library. DNA sequences flanking the sequence (TG)₁₄TTGGGAATGGGGCTGGGGGGAGAC-AGGGT(TG)₁₀ within HaeIII subclone 2E12B of this cosmid (EMBL accession number X56973) were used to design PCR primers.

PCR Primers: 2E12B #1: 5'-TGGAACACTCAGGCGA-3'
2E12B #2: 5'-CCAGAGCCACTTTCTAC-3'

Polymorphism: Ten allelic fragments were resolved on DNA sequencing gels. Lengths of allelic fragments (nt) were: A1 = 193, A2 = 191, A3 = 189, A4 = 187, A5 = 185, A6 = 183, A7 = 181, A8 = 161, A9 = 159, A10 = 155.

Frequencies: Allele frequencies in 52 unrelated Caucasians were: A1 = .01, A2 = .01, A3 = .11, A4 = .13, A5 = .26, A6 = .02, A7 = .06, A8 = .01, A9 = .38, A10 = .01; PIC = 0.72.

Chromosomal Localization and Mendelian Inheritance: PCR of genomic DNAs from two somatic cell hybrid panels indicated localization to chromosome 14. Fluorescent in situ hybridization indicated localization to 14q24.3. Linkage analysis in 8 CEPH families with 85 informative meioses against locus D14S24 (probe CRI-C70) gave a maximum LOD score of 2.95 at $\theta = 0.3$. Mendelian inheritance was observed in all cases.

PCR Conditions: PCR reactions are carried out in a total volume of 25 μ l containing: 25 ng genomic DNA, 10 pmole of each primer, 1.5 mM MgCl₂, 200 μ M dNTPs, 50 mM KCl, 10 mM Tris-Cl, pH 8.3, 0.6 units Taq polymerase (Ampli-Taq) and 0.01% gelatin. Amplification is for 35 cycles with denaturation at 94°C for 60 seconds, annealing at 54°C for 60 seconds and extension at 72°C for 30 seconds.

Comments: This marker locus will be a useful index marker (1) for the genetic linkage map of chromosome 14.

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Reference: 1) Roberts,L. (1990) *Science* **248**, 805.

A PstI DNA polymorphism in the human stefin A gene (STF 1)

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Source/Description: Probe STFA242 (1) containing ~1500 bp EcoRI-BamHI DNA fragment including the stefin A coding region.

Polymorphism: A two-allele polymorphism with fragments of 10.2 kb (A) and 9.4 kb (B) is identified after hybridization with PstI digested human genomic DNA.

Not Polymorphic For: EcoRI, HindIII and TaqI.

Frequency: 60 unrelated Caucasians were studied.

A1 allele: 0.55

A2 allele: 0.45

Chromosomal Location: The stefin A has been mapped to chromosome 3cen-q21 (2).

Mendelian Inheritance: Codominant segregation has been demonstrated in 8 families.

Other Comments: High stringency wash of the blots in 0.1 $\times$ SSC/0.1% SDS at 65°C shows no significant background. A constant band at 1.5 kb was observed.

Probe Availability: Available on request from Wang-Ting Hsieh or Dunne Fong.

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References: 1) Kartasova,T. *et al.* (1987) *Nucl. Acids Res.* **15**, 5945-5962. 2) Hsieh,W.-T. *et al.* (1991) *Genomics* **9**, 207-209.

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