Dinucleotide repeat polymorphism at the D14S42 locus

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Source/Description: A genomic *Sau3A* fragment isolated from a *HindIII* 4.0 kb insert in a human chromosome 14 specific library (LL14NS01) was cloned into pUC19 and selected by hybridisation to poly(dC-dA) poly(dG-dT). The clone was designated SJ10F. Sequencing of SJ10F provided the information necessary to synthesise two oligonucleotides homologous to the regions flanking the tandem repeat which were used to amplify a fragment using Polymerase Chain Reaction. The predicted length of the amplified region was 123 bp.

Primer Sequences:

GGTCACTGATCGCCTTTGAA—CA strand

GGAGGTTGTGAACACCTTGA—GT strand

Frequency: Estimated from 154 chromosomes of unrelated CEPH family parents (Caucasians) and 26 chromosomes of unrelated Irish individuals. PIC = 0.65.

Allele (bp)	Frequency	Allele (bp)	Frequency
133	0.0222	123	0.1333
131	0.0612	121	0.5333
129	0.0945	119	0.0111
127	0.0444	117	0.0111
125	0.0722	115	0.0167

Chromosomal Location: The results of amplification using DNA templates isolated from panels of somatic cell hybrids (BIOS) were consistent with assignment of the dinucleotide repeat to Chromosome 14.

Mendelian Inheritance: Co-dominant segregation was observed in one large two generation Irish family TCDG1 and in 40 CEPH families.

Other Comments: The PCR reactions were carried out as described in the reference (1) except that 50 mM KCl was replaced by 10 mM KCl, 10 mM NH₄Cl. The amplification conditions were: 94°C; 7 min, followed by 30 cycles of 94°C; 1 min, 55°C; 2 min, 72°C; 3 min. The PCR product was fractionated on 8% denaturing polyacrylamide gels. The allele sizes were calculated by comparison to pUC19 DNA sequencing ladder. The dinucleotide repeat sequence in SJ10F was of the form CT(AC)₁₈AA. The sequence has been submitted to Genbank, accession number X54720.

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Reference: Weber, J.L. and May, P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396.

Dinucleotide repeat polymorphism at the D6S109 locus

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Source/Description: A 6 kb human genomic fragment was generated from a human-hamster radiation hybrid cell line (R78) containing a small portion of human chromosome 6p (Zoghbi *et al.* in press) by Alu-PCR (Nelson *et al.*, 1989). Primer 15a is from the 3' region of one of the end Alu-repeat sequences, and primer 15b is located on the other side of the dinucleotide repeat.

PCR Primers:

15a 5' CAC CCT GGG CAA TAA GAG CG 3' (GT strand)

15b 5' CCC CTC TTC ATC CTC CCT TTC A 3' (CA strand)

Frequency: Estimated from 174 chromosomes of unrelated CEPH family grandparents (Caucasians). PIC = 0.78.

Allele (bp)	Frequency	Allele (bp)	Frequency
A1 193	0.02	A6 183	0.07
A2 191	0.13	A7 181	0.07
A3 189	0.17	A8 175	0.03
A4 187	0.37	A9 169	0.01
A5 185	0.13		

Chromosomal Localization: Assigned to chromosome 6p between HLA-A and F13a using a panel of human-hamster radiation hybrids containing 6p fragments.

Mendelian Inheritance: Co-dominant segregation was observed in 40 CEPH reference families.

Other Comments: Conditions for the amplification reactions were as described by the United States Biochemical Corporation except that 2.0 mM MgCl₂ was used, 1/10 the amount of the repetitive Alu-primer (15a) was added and samples were processed through 25 temperature cycles consisting of 1 min at 94°, 1 min at 60°, and 45 sec at 72°. Before PCR amplification, the 5' end of the 15b primer was labelled with γ S³⁵-ATP. The dinucleotide repeat sequence in pLR15 was of the form (CA)₂₃.

Probe Availability: pLR15 is freely available. Contact Harry T.Orr.

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References: 1) Nelson, D.L., Ledbetter, S.A., Corbo, L., Victoria, M.F., Ramirez-Solis, R., Webster, T.D., Ledbetter, D.H. and Caskey, C.T. (1989) *Proc. Natl. Acad. Sci. USA* **86**, 6686–6690. 2) Zoghbi, H.Y., McCall, A.E., LeBorgne-Demarquoi, F. *Genomics*, (in press).

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