Dinucleotide repeat polymorphism at the D19S49 locus

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Source/Description: A genomic *Sau3A* I fragment from a large insert human chromosome 19-specific phage library (LL19NL01) was cloned into mp18 and selected by hybridization to poly(dC-dA)·poly(dG-dT). The cloned fragment was designated Mfd11. Sequencing of Mfd11 provided the information necessary for polymerase chain reaction primer synthesis. The clone length was 265 bp, and the predicted length of the amplified fragment was 121 bp.

Primer Sequences: ACTCATGAAGGTGACAGTTC (CA strand); GTGTTGTTGACCTATTGCAT (GT strand)

Frequency: Estimated from 152 chromosomes of unrelated CEPH family parents (Caucasians). PIC = 0.71.

Allele(bp)	Frequency	Allele(bp)	Frequency
122	0.07	114	0.05
120	0.16	110	0.01
118	0.17	108	0.43
116	0.10	106	0.01

Chromosomal Localization: Assignment to chromosome 19 was verified using DNA templates isolated from panels of somatic cell hybrids.

Mendelian Inheritance: Co-dominant segregation was observed in 40 two or three generation families.

Other Comments: Conditions for the amplification reactions were as described in the reference except that samples were processed through 27 temperature cycles consisting of 1 min at 94°, 2 min at 55° and 2 min at 72°. Sizes of the alleles were determined by comparison to mp8 DNA sequencing ladders and were the averages of the sizes of the CA and GT strand bands. The dinucleotide repeat sequence in Mfd11 was of the form (AC)₂₃A. The sequence of Mfd11 has been submitted to GenBank.

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

The anonymous probe DR281 (D1S99) identifies a *MspI* polymorphism

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Source and Description: DR281 is a 1.2 kb human DNA fragment subcloned into the *PstI* site of pBR322.

Polymorphism: *MspI* identifies a three allele polymorphism with fragments of 4.4 kb, 4.2 kb and 3.6 kb.

Frequency: Studied in 42 unrelated caucasians.

4.4 kb allele 0.25

4.2 kb allele 0.30

3.6 kb allele 0.45

Not Polymorphic For: *EcoRI*, *EcoRV*, *BamHI*, *HindIII*, *BglII*, *TaqI* and *SstI* with DNA from 9 unrelated caucasians.

Chromosomal Localization: The polymorphism has been localised to 1q21–1qter using a panel of somatic cell hybrids.

Mendelian Inheritance: Co-dominant segregation shown in 2 informative families (20 individuals) (see figure).

Probe Availability: On request from A.J. Driesel.

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