Dinucleotide repeat polymorphism at the D9S116 locus (9q31 – 34)

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Source/Description: A human genomic cosmid, 6E5B (31/32), was isolated from a cosmid library made from a cell line, PK87–9, that contains human chromosome 9 as its sole human component (1). Flanking sequence for a GT repeat is Genbank X62924.

Primer Sequences: GTCCATTTCTCTAACACCCG (GT strand), and TACCCTAGAGCCTGGATGG (CA strand).

Frequency: Estimated from 82 chromosomes of unrelated North Americans and members of the Venezuelan reference pedigree. Heterozygosity = 0.85. Genotype of CEPH parents: 10201- A1, A3; 10401- A5, A7.

Allele	bp	Frequency	Allele	bp	Frequency
A1	112	.01	A7	100	.23
A2	110	.01	A8	98	.10
A3	108	.17	A9	96	.03
A4	106	.12	A10	90	.01
A5	104	.14	A11	88	.01
A6	102	.17			

Chromosomal Localization: D9S116 has been localized to chromosome 9q31–34 by somatic cell hybrid mapping (1) and linkage mapping.

Mendelian Inheritance: Co-dominant inheritance was observed in five two-generation families.

PCR Conditions/Comments: PCR was performed as described (1), with 29 cycles of denaturation at 94°C for 30 s, annealing at 58°C for 30 s, and extension at 72°C for 40 s in 1.5 mM Mg²⁺. Internal labelling was used, and allele sizes were determined by polyacrylamide gel electrophoresis with 7 M urea.

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Reference: 1) Kwiatkowski,D.J. *et al.* (1992) *Genomics* **12**, 229–240.

Dinucleotide repeat polymorphism at the D9S117 locus (9q31 – 34)

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Source/Description: A human genomic cosmid, 6E11B (18/19), was isolated from a cosmid library made from a cell line, PK87–9, that contains human chromosome 9 as its sole human component (1). Flanking sequence for a GT repeat is Genbank X62925.

Primer Sequences: GGTGCAATTAGAGGTGGTGA (GT strand), and AAAATACACGCAGAGACGTAG (CA strand).

Frequency: Estimated from 80 chromosomes of unrelated North Americans and members of the Venezuelan reference pedigree. Heterozygosity = 0.78. Genotype of CEPH parents: 10201- A5, A6; 10401- A5, A6.

Allele	bp	Frequency	Allele	bp	Frequency
A1	120	.01	A5	112	.25
A2	118	.08	A6	110	.09
A3	116	.15	A7	108	.09
A4	114	.31	A8	106	.02

Chromosomal Localization: D9S117 has been localized to chromosome 9q31–34 by somatic cell hybrid mapping (1) and linkage mapping.

Mendelian Inheritance: Co-dominant inheritance was observed in five two-generation families.

PCR Conditions/Comments: PCR was performed as described (1), with 29 cycles of denaturation at 94°C for 30 s, annealing at 58°C for 30 s, and extension at 72°C for 40 s in 1.5 mM Mg²⁺. Internal labelling was used, and allele sizes were determined by polyacrylamide gel electrophoresis with 7 M urea.

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