Dinucleotide repeat polymorphism at the D9S120 locus (9q31 – 34)

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Source/Description: A human genomic cosmid, 9D5A (12/94), 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 X62927.

Primer Sequences: GCAGCCCAACACAAATTCATA (GT strand), and AGAACTGTCTTGGGCCACC (CA strand).

Frequency: Estimated from 82 chromosomes of unrelated North Americans and members of the Venezuelan reference pedigree. Heterozygosity = 0.76. Genotype of CEPH parents: 10201- A3, A4; 10401- A3, A4.

Allele	bp	Frequency	Allele	bp	Frequency
A1	155	.03	A5	147	.12
A2	153	.12	A6	145	.13
A3	151	.21	A7	143	.06
A4	149	.32	A8	141	.01

Chromosomal Localization: D9S120 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 D9S121 locus (9q31 – 34)

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Source/Description: A human genomic cosmid, 10G1A (33/43), 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 X62928.

Primer Sequences: CGTAGCGTGGAGTAGCTCT (GT strand), and GGCAATAGAGTGAGACTCCA (CA strand).

Frequency: Estimated from 86 chromosomes of unrelated North Americans and members of the Venezuelan reference pedigree. Heterozygosity = 0.79. Genotype of CEPH parents: 10201- A3, A5; 10401- A4, A4.

Allele	bp	Frequency	Allele	bp	Frequency
A1	142	.04	A6	132	.03
A2	140	.15	A7	130	.03
A3	138	.13	A8	128	.05
A4	136	.40	A9	126	.01
A5	134	.16			

Chromosomal Localization: D9S121 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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