Dinucleotide repeat polymorphism at the D9S123 locus (9q31 – 34)

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Source/Description: A human genomic cosmid, 1AE6A (65/66), 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 X62929.

Primer Sequences: GTGAAGGGACCCAGAACTC (GT strand), and CCCTTCTGCATTTCTGCATG (CA strand).

Frequency: Estimated from 80 chromosomes of unrelated North Americans and members of the Venezuelan reference pedigree. Heterozygosity = 0.55. Genotype of CEPH parents: 10201- A1, A7; 10401- A7, A7.

Allele	bp	Frequency	Allele	bp	Frequency
A1	92	.10	A6	80	.09
A2	88	.03	A7	78	.66
A3	86	.02	A8	76	.05
A4	84	.01	A9	74	.01
A5	82	.03			

Chromosomal Localization: D9S123 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.

Acknowledgements: This work was supported by NIH grants HG00324 and HG00169, the Dystonia Medical Research Foundation, and the National Tuberous Sclerosis Association.

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