Dinucleotide repeat polymorphism at the GSN locus (9q32-34)

D.J.Kwiatkowski and S.Perman
Massachusetts General Hospital, Hematology-Oncology Unit, Bldg. 149, 13th St., 8 West, Charlestown, MA 02129, USA

Source/Description: A human genomic 1.5 kb EcoRI-PstI fragment from an intron in the 3′ region of the GSN gene was selected by hybridization to poly(dC-dA)·poly(dG-dT). The cloned fragment was designated D3d. Sequencing was performed for determination of primer sequences for the polymerase chain reaction, which was predicted to amplify a sequence of length 129 bp.

Primer Sequences: CAGCCAGCTTTGGAGACAAC (CA strand); TCGCAAGCATATGACTGTAA (GT strand).

Frequency: Estimated from 66 chromosomes of unrelated North Americans PIC = 0.76, heterozygosity = 0.70.

allele (bp)	frequency	allele (bp)	frequency
D1 147	0.02	D7 131	0.01
D2 145	0.02	D8 129	0.06
D3 143	0.17	D9 127	0.40
D4 141	0.05	D10 125	0.05
D5 139	0.02	D11 113	0.01
D6 127	0.20	D12 111	0.01

Chromosomal Localization: GSN has been localized to chromosome 9q32-34[1].

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

PCR Conditions/Comments: PCR conditions were as described[2], with 25 cycles of denaturation at 94° for 1 min, annealing at 55° for 1 min, and extension at 72° for 1 min. Sizes of alleles were determined by denaturing polyacrylamide gel electrophoresis and autoradiography with comparison to the cloned sequence and standard DNA samples. The core repeat was $(AC)_{16}A_3(CA)_3$. The sequence has been submitted to Genbank.

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References: 1) Kwiatkowski,D.J. *et al.* (1988) *Am. J. Hum. Genet.* **42**, 565-572. 2) Kwiatkowski,D.J. *et al.* (1991) *Am. J. Hum. Genet.* **48**, 121-128.

Dinucleotide repeat polymorphism at the ABL locus (9q34)

D.J.Kwiatkowski
Massachusetts General Hospital, Hematology-Oncology Unit, Bldg. 149, 13th St., 8 West, Charlestown, MA 02129, USA

Source/Description: A human genomic 2.0 kb EcoRI fragment from the intron between exons 1a and 2 in the c-abl gene[1] (provided by D. Leibowtiz) was selected by hybridization to poly(dC-dA)·poly(dG-dT). The cloned fragment was designated 2D. Sequencing was performed for determination of primer sequences for the polymerase chain reaction, which was predicted to amplify a sequence of length 105 or 141 bp.

Primer Sequences: TTTACACCTTCACCCAGAGA (CA strand); GGCTGTGTTCAGTTAAACGT or CTGACAC-AGTGTTTTGGGAT (GT strand, 105 and 141 bp, respectively).

Frequency: Estimated from 55 chromosomes of unrelated North Americans. PIC = 0.64, heterozygosity = 0.67.

allele (bp)	frequency	allele (bp)	frequency
F1 117	0.01	F6 103	0.06
F2 113	0.01	F7 101	0.13
F3 111	0.01	F8 93	0.01
F4 107	0.26	F9 89	0.03
F5 105	0.48		

Chromosomal Localization: ABL has been localized to chromosome 9q34[1].

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

PCR Conditions/Comments: PCR conditions were as described[2], with 25 cycles of denaturation at 94° for 1 min, annealing at 55° for 1 min, and extension at 72° for 1 min. Sizes of alleles were determined by denaturing polyacrylamide gel electrophoresis with autoradiography with comparison to the cloned sequence and standard DNA samples. The core repeat was $(TG)_1CT(TG)_7$. The sequence of 2D has been submitted to Genbank.

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References: 1) Leibowtiz,D. *et al.* (1985) *Blood* **65**, 526-529. 2) Kwiatkowski,D.J. *et al.* (1991) *Am. J. Hum. Genet.* **48**, 121-128.