

Dinucleotide repeat polymorphism at the D21S145 locus

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Source/Description: pMC1.44g is a 0.4 kb EcoRI/HindIII subclone of phage fVC1.44, isolated from an EMBL4 human chromosome 21 library (1). The sequence of pMC1.44g contains a (CA)₁₅ repeat (EMBL accession no. X63572).

PCR Primers:

P1.44-1: 5'-CTT CTC TTG ATT GTG TGT GT-3'

P1.44-2: 5'-AAC ATA TCT CTG AAT ATC GG-3'

Polymorphism: 6 alleles were observed in 46 unrelated Caucasians.

Allele	Length	Frequency	Allele	Length	Frequency
A1	180 bp	.239	A4	174 bp	.152
A2	178 bp	.511	A5	172 bp	.076
A3	176 bp	.011	A6	168 bp	.011

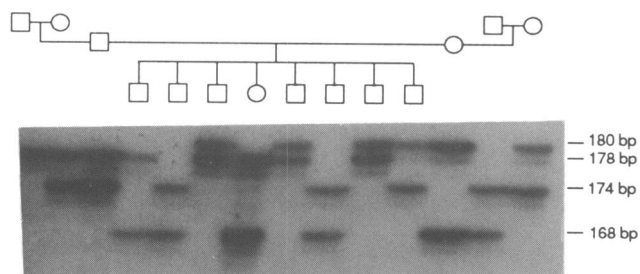
PIC = 0.60.

Chromosomal Location: fVC1.44 (D21S145) is located on chromosome 21, in 21q21.1-q21.2 (2).

Mendelian Inheritance: Co-dominant inheritance was demonstrated in CEPH families 1333, 1334 and 1347.

PCR Conditions: The PCR reaction is carried out in a total volume of 25 μ l containing approximately 200 ng genomic DNA, 1 unit Taq DNA polymerase, 25 pmol of each primer, 0.4 pmol γ -³²P end-labelled primer P1.44-1, 200 μ M dNTP's, 10 mM Tris-HCl pH 8.3, 50 mM KCl, 1.5 mM MgCl₂, 0.01% gelatin. Amplification is for 22 cycles with denaturation at 94°C for 60 seconds, annealing at 54°C for 90 seconds and extension at 72°C for 120 seconds. Aliquots of the PCR products are denatured, separated on a DNA sequencing gel and autoradiographed.

References: 1) Van Camp *et al.* (1990) *Som. Cell Mol. Genet.* **16**, 241-249. 2) Van Camp *et al.* (1991) *Hum. Genet.* **87**, 649-653.



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Dinucleotide repeat polymorphism at the D1S16 locus

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Source/Description: pMCS16.1 is a 1.2 kb Sau3A subclone of cosmid ICRFc102B05120, isolated from a flow-sorted human chromosome 21 cosmid library after screening with probe pGSE9 (1, 2). The sequence of pMCS16.1 contains a (CA)₂₀ repeat (EMBL accession number X63573).

PCR Primers:

PS16.5: 5'-TCA TTT ACT TTG GAA GTC AAT ATT C-3'

PS16.6: 5'-ACA ACA GTA AAC CAG CTT ATT ATT C-3'

Polymorphism: 8 alleles were observed in 80 unrelated Caucasians:

Allele	Length	Frequency	Allele	Length	Frequency
C1	175 bp	.031	C5	167 bp	.019
C2	173 bp	.056	C6	165 bp	.006
C3	171 bp	.231	C7	155 bp	.006
C4	169 bp	.175	C8	153 bp	.475

PIC = 0.64

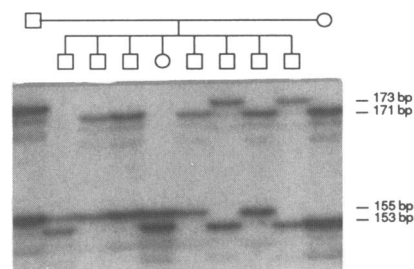
Chromosomal Location: pGSE9 (D21S16) is located on chromosome 21, in 21q11.1 (3).

Mendelian Inheritance: Co-dominant inheritance was demonstrated in CEPH families 1333, 1334 and 1347.

PCR Conditions: The PCR reaction is carried out in a total volume of 25 μ l containing approximately 200 ng genomic DNA, 1 unit Taq DNA polymerase, 25 pmol of each primer, 0.4 pmol γ -³²P end-labelled primer PS16.5, 200 μ M dNTP's, 10 mM Tris-HCl pH 8.3, 50 mM KCl, 1.5 mM MgCl₂, 0.01% gelatin. Amplification is for 22 cycles with denaturation at 94°C for 60 seconds, annealing at 55°C for 90 seconds and extension at 72°C for 120 seconds. Aliquots of the PCR products are denatured, separated on a DNA sequencing gel and autoradiographed.

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References: 1) Stewart *et al.* (1985) *Nucl. Acids Res.* **13**, 4125-4132. 2) Nizetic *et al.* (1991) *Proc. Natl. Acad. Sci. USA* **88**, 3233-3237. 3) Gardiner *et al.* (1990) *EMBO J.* **9**, 25-34.



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