Dinucleotide repeat polymorphism at the D21S145 locus

M.Cruts¹, H.Backhovens¹,² and C.Van Broeckhoven¹*
¹University of Antwerp, UIA, Department of Biochemistry, Laboratory of Neurogenetics, Born-Bunge Foundation, Universiteitsplein 1, B-2610 Antwerpen and ²Innogenetics Inc., Industriepark Zwijnaarde, B-9710 Gent, Belgium

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.

<table>
<thead>
<tr>
<th>Allele</th>
<th>Length</th>
<th>Frequency</th>
<th>Allele</th>
<th>Length</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1</td>
<td>180 bp</td>
<td>.239</td>
<td>A4</td>
<td>174 bp</td>
<td>.152</td>
</tr>
<tr>
<td>A2</td>
<td>178 bp</td>
<td>.511</td>
<td>A5</td>
<td>172 bp</td>
<td>.076</td>
</tr>
<tr>
<td>A3</td>
<td>176 bp</td>
<td>.011</td>
<td>A6</td>
<td>168 bp</td>
<td>.011</td>
</tr>
</tbody>
</table>
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 μl containing approximately 200 ng genomic DNA, 1 unit Taq DNA polymerase, 25 pmol of each primer, 0.4 pmol γ³²P end-labelled primer P1.44-1, 200 μM 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.


* To whom correspondence should be addressed

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Dinucleotide repeat polymorphism at the D1S16 locus

M.Cruts¹, H.Backhovens¹,² and C.Van Broeckhoven¹*
¹University of Antwerp, UIA, Department of Biochemistry, Laboratory of Neurogenetics, Born-Bunge Foundation, Universiteitsplein 1, B-2610 Antwerpen and ²Innogenetics Inc., Industriepark Zwijnaarde, B-9710 Gent, Belgium

Source/Description: pMC16.1 is a 1.2 kb Sau3A subclone of cosmid ICRFc102B05120, isolated from a flow-sorted human chromosome 21 library after screening with probe pGSE9 (1, 2). The sequence of pMC16.1 contains a (CA)₂₀ repeat (EMBL accession number X63573).

PCR Primers:
PS16.5: 5'-TCA TTT ACT TTG GAA GTC AAT ATT C-3'
PS 16.6: 5'-ACA ACA GTA AAC CAG CTT ATT ATT C-3'

Polymorphism: 8 alleles were observed in 80 unrelated Caucasians:

<table>
<thead>
<tr>
<th>Allele</th>
<th>Length</th>
<th>Frequency</th>
<th>Allele</th>
<th>Length</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>C1</td>
<td>175 bp</td>
<td>.031</td>
<td>C5</td>
<td>167 bp</td>
<td>.019</td>
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<tr>
<td>C2</td>
<td>173 bp</td>
<td>.056</td>
<td>C6</td>
<td>165 bp</td>
<td>.006</td>
</tr>
<tr>
<td>C3</td>
<td>171 bp</td>
<td>.231</td>
<td>C7</td>
<td>155 bp</td>
<td>.006</td>
</tr>
<tr>
<td>C4</td>
<td>169 bp</td>
<td>.175</td>
<td>C8</td>
<td>153 bp</td>
<td>.475</td>
</tr>
</tbody>
</table>
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 μl containing approximately 200 ng genomic DNA, 1 unit Taq DNA polymerase, 25 pmol of each primer, 0.4 pmol γ³²P end-labelled primer PS16.5, 200 μM 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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* To whom correspondence should be addressed