A dinucleotide repeat polymorphism at the D1S116 locus

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Source and Description: Cosmid ICRFc102D0922 was from a flow sorted chromosome 21 cosmid library. DNA sequences flanking a (GT)₁₇ repeat within an AluI subclone (2D9-1) of this cosmid (EMBL accession # X54557) were used to design PCR primers.

PCR Primers:

2D9 #1-5'-TACAAGGCAACCACATAATT-3'

2D9 #2-5'-CTTTCCTAATTGTGTGTGT-3'

Polymorphism: Allelic fragments were resolved on DNA sequencing gels. Lengths of allelic fragments (nt) were: A1 = 101, A2 = 99, A3 = 97, A4 = 95, A5 = 93, A6 = 91, A7 = 89.

Frequencies: Allele frequencies in 39 unrelated European Caucasians were:

A1 = .077

A2 = .103

A3 = .217

A4 = .013

A5 = .013

A6 = .051

A7 = .525

PIC = 0.62

Chromosomal Localization and Mendelian Inheritance: Fluorescence in situ hybridization localized cosmid ICRFc102D0922 to 1p31.2. Linkage analysis in 2 CEPH families showed no recombination in 14 informative meioses with D1S22 (localized to pter-p22 (1)), giving a maximum LOD score of 4.50 at theta = 0. Mendelian inheritance was observed in all cases.

PCR Conditions: We carry out PCR in a total volume of 25 µl containing: 25 ng genomic DNA, 10 pmole of each primer, 1.5 mM MgCl₂, 200 µM dNTPs, 50 mM KCl, 10 mM tris-Cl, pH 8.3, 0.6 units Taq polymerase (BRL) and 0.01% gelatin. Amplification is for 35 cycles with denaturation at 94°C for 30 seconds, annealing at 46°C for 45 seconds and extension at 72°C for 1.5 minutes.

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Reference: 1) Bruns, G.A.P. and Sherman, S.L. (1989) *Cytogenet. Cell Genet.* 51, 67-90.

Dinucleotide repeat polymorphism at the D21S172 locus

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Source and Description: Cosmid ICRFc102H0921 was from a flow sorted chromosome 21 cosmid library. DNA sequence flanking a (CA)₂₀ repeat within a Sau3A subclone (1H9) of this cosmid (EMBL accession # X56513) were used to design PCR primers.

PCR Primers:

1H9 #2: 5'-TACAGTGGCAAATGTCATTG-3'

1H9 #3: 5'-GAATATGTGTTAGGTCCTGC-3'

Polymorphism: Eight allelic fragments were resolved on DNA sequencing gels. Lengths of the allelic fragments (nt) were: A1 = 161, A2 = 157, A3 = 155, A4 = 153, A5 = 151, A6 = 149, A7 = 147, A8 = 145.

Frequencies: Allele frequencies in 51 unrelated European Caucasians were:

A1 = .01

A2 = .029

A3 = .098

A4 = .049

A5 = .588

A6 = .176

A7 = .02

A8 = .029

PIC = .58

Chromosomal Localization and Mendelian Inheritance: Fluorescence in situ hybridization localized cosmid ICRFc102H0921 to 21q11.2. Linkage with 4 CEPH families showed no recombination in 35 informative meioses with D21S13 probe D21K9 (localized to q11.2), giving a maximum LOD score of 9.62 at theta = 0. Mendelian inheritance was observed in all cases.

PCR Conditions: PCR reactions are carried out in a total volume of 25 µl containing: 25 ng genomic DNA, 10 pmole of each primer, 1.5 mM MgCl₂, 200 µM dNTPs, 50 mM KCl, 10 mM tris-Cl, pH 8.3, 0.6 units Taq polymerase (Cetus) and 0.01% gelatin. Amplification is for 35 cycles with denaturation at 94°C for 45 seconds, annealing at 56°C for 45 seconds and extension at 72°C for 45 seconds.

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