

Polymorphic dinucleotide repeats at the D3S1417, D3S1418 and D12S271 loci

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Source/Description: A plasmid library of flow-sorted human chromosome 21 (1) was screened with a (GT)₁₀ oligonucleotide as described (2). Five of 17 positive clones did not map to chromosome 21. Three of these five clones contain highly polymorphic loci and are designated GT01 (D3S1417), GT06 (D3S1418) and GT22 (D12S271).

Primer Sequences: GT01 (D3S1417)

Forward 5' CTC ACT GCC AGC TCT GTC AA 3'

Reverse 5' TTT (T/A)AA ATA CCA TGG GTT AG 3'

The dinucleotide repeat sequence was of the form (GT)₁₀Allele Frequency: D3S1417

Estimated from 20 CEPH unrelated individuals. Observed heterozygosity was 0.65 (13 of 20)

Allele	(bp)	Frequency
1	220	0.03
2	218	0.18
3	216	0.2
4	214	0.33
5	212	0.15
6	210	0.08
7	208	0.05

Chromosome Localization: Locus D3S1417 was mapped to chromosome 3 by PCR amplification of the somatic cell hybrids of NIGMS, panel 2 (3). Linkage analysis using genotypes from three informative CEPH pedigrees (1329, 1331, 1349) against the CEPH V6 database showed closest linkage of D3S1417 to D3S1282 with a lod score of 5.42 at a theta of 0.000.

Primer Sequences: GT06 (D3S1418)

Forward 5' CAA GGT TAT GCA ATA ATA GT 3'

Reverse 5' TCT AAC TCA AAA GAA GTC AC 3'

The dinucleotide repeat sequence was of the form (GT)₁₃(A-G)₁₁Allele Frequency: D3S1418

Estimated from 32 CEPH unrelated individuals. Observed heterozygosity was 0.84 (26 of 31)

Allele	(bp)	Frequency
1	242	0.02
2	240	0.03
3	236	0.08
4	234	0.13
5	232	0.19
6	230	0.06

7	228	0.17
8	226	0.02
9	224	0.05
10	222	0.27

Chromosome Localization: Locus D3S1418 was mapped on chromosome 3 by PCR amplification of somatic cell hybrids of NIGMS panel 2 (3). Linkage analysis using genotypes from three informative CEPH pedigrees (12, 1349, 1421) against the CEPH V6 database showed closest linkage of D3S1418 to D3S196 (Mfd 17) with a lod score of 11.74 at a theta of 0.00. Sequence and genotype comparisons revealed that the two loci are not identical.

Primer Sequences: GT22 (D12S271)

Forward 5' CTT TGC AAA TCA CGT ATC TG 3'

Reverse 5' ACC ATC CCA GTT GAA CAG TG 3'

The dinucleotide repeat sequence was of the form (GT)₁₄Allele frequency: D12S271

Estimated from 23 unrelated CEPH individuals. Observed heterozygosity was 0.65 (15 of 23)

Allele	(bp)	Frequency
1	102	0.07
2	100	0.59
3	98	0.32
4	96	0.02

Chromosome Localization: Locus D12S271 was mapped on chromosome 12 by PCR amplification of somatic cell hybrids of NIGMS panel 2 (3) and the BIOS PCRable panel (4). Linkage analysis using genotypes from four informative CEPH pedigrees (12, 1345, 1413, 1421) against the CEPH V6 database showed closest linkage of D12S271 to D12S28 (CRI-L375) with a lod score of 3.61 at a theta of 0.00.

PCR conditions were described in ref. 2.

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