Dinucleotide repeat polymorphism at the D17S513 locus

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Source and Description of Clone: A chromosome 17- specific Ch40 lambda phage library created at Los Alamos (van Dilla *et al.*, 1986) was restricted with BamHI and subcloned into pTZ18R. The clone was selected by hybridization to poly (dC-dA)-poly(dG-dT). Sequencing of the cloned fragment, 12G6, provided data for polymerase chain reaction primer synthesis. The 193 bp amplified fragment contained a (AC)₂₃ dinucleotide repeat sequence.

PCR Primer Sequences:

12G6 (CA strand) 5'-TTCACCTGTGGGCTGCTGTC-3'
12G6 (TG strand) 5'-TAAGAAAGGCTCCCAAGCA-3'

Polymorphism: Frequencies of the alleles of the dinucleotide repeat polymorphism (Weber and May, 1989) were estimated from 36 chromosomes of unrelated CEPH family members (Caucasians). Observed heterozygosity was 0.89 and expected heterozygosity was 0.74.

Allele (bp)	Frequency	Allele (bp)	Frequency
A1 183	0.36	A5 193	0.33
A2 185	0.06	A6 195	0.11
A3 189	0.06	A7 199	0.03
A4 191	0.03	A8 203	0.03

Chromosomal Localization: The sequence was localized by genetic analysis of key recombinants in the CEPH public database (Dausset, 1986) to chromosome 17 in the region between D17S126 and MYH2 and therefore to 17p13.

Mendelian Inheritance: Co-dominant segregation was observed in 5 three generation CEPH families.

Other Comments: The PCR reaction was performed on 30 ng of genomic DNA using 3 pmoles of each oligonucleotide primer. The samples were processed as described (Weber and May, 1989) except that dCTP was 5 μ M with 2.5 μ M Ci ³²P. dCTP at 6000Ci/mmol. Samples were cycled 27 times at 94°C for 1 min, 55°C for 2 min and 72°C for 1 min.

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References: 1)Dausset,J. (1986) *La Press Medical* **15**, 1801–1802. 2) van Dilla,M.A. *et al.* (1986) *Biotechnology* **4**, 537–552. 3) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396.

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Source and Description of Clone: A chromosome 17-specific Ch40 lambda phase library created at Los Alamos (2) was restricted with BamHI and subcloned into pTZ18R. The clone was selected by hybridization to poly(dC-dA)-poly(dG-dT). Sequencing of the cloned fragment, 26C2, provided data for polymerase chain reaction primer synthesis. The 129 bp fragment contained a (CA)₂₁ dinucleotide repeat sequence.

PCR Primer Sequences:

26C2 (CA strand) 5'-ATTAAGAGCAGAACTCTCTTTCC-3'
26C2 (TG strand) 5'-ATTTCTAAAGAACTTTTG-
ATAAGCA-3'

Polymorphism: Frequencies of the alleles of the dinucleotide repeat polymorphism (3) were estimated from 38 chromosomes of unrelated CEPH family members (Caucasians). The observed heterozygosity was 0.53 and the expected heterozygosity was 0.57.

Allele	(bp)	Frequency	Allele	(bp)	Frequency
A1	129	0.23	A4	123	0.05
A2	127	0.60	A5	119	0.03
A3	125	0.08			

Chromosomal Localization: The sequence was localized by genetic analysis of key recombinants in the CEPH public database (1) to chromosome 17 in the region between D17S71 and HOX2 and therefore between 17p11.2 and 17q22.

Mendelian Inheritance: Co-dominant segregation was observed in 5 three-generation CEPH families.

Other Comments: The PCR reaction was performed on 30 ng of genomic DNA using 10 pmoles of each oligonucleotide primer. The samples were processed as described (3) except that dCTP was 5 μ M with 2.5 μ M Ci ³²P-dCTP at 6000 Ci/mmol. Samples were cycled 27 times at 94°C for 1 min, 55°C for 1.5 min and 72°C for 1.5 min. The dinucleotide repeat was based on a (C-A)₂₁ sequence.

Acknowledgement: This work was supported by grant CA48711.

References: Dausset,J. (1986) *La Press Medical* **15**, 1801–1802. 2) van Dilla,M.A. *et al.* (1986) *Biotechnology* **4**, 537–552. 3) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396.