

Dinucleotide repeat polymorphism at the human Poly (ADP-Ribose) polymerase gene (PPOL)

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Source/Description: The polymorphism is a dinucleotide repeat of the form (AC) 12A and starts at position 845 (exon 1) of the human Poly (ADP-Ribose) Polymerase gene on chromosome 1q41-q42 (Genbank accession no. M34337) (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as previously described (2). The predicted size of the amplified sequence is 85 bp.

Primer Sequence:

GATCCCCATCTCTTTCTTT (CA strand)
AAATTGTGGTAATGACTGCA (TG strand)

Frequency: Estimated from 110 chromosomes of unrelated CEPH individuals. Observed heterozygosity is 63% with a PIC of 0.58.

Allele	(bp)	Frequency
C1	99	0.026
C2	97	0.130
C3	93	0.050
C4	93	0.245
C5	87	0.009
C6	85	0.540

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

Chromosomal Localization: The human PPOL gene has been assigned to chromosome 1q41-q42 (3).

PCR Conditions: The PCR reaction was performed using 200 ng genomic DNA, 10 pMoles of each primer and 0.75 units of Taq polymerase in a final volume of 50 μ l. Samples were subjected to 30 cycles consisting of 40 seconds at 92°C, 30 seconds at 50°C and 30 seconds at 72°C.

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References: 1) Ogura, T. *et al.* (1990) *Biochem. Biophys. Res. Commun.* **167**, 701–710. 2) Weber, J.L. and May, P.E. (1989) *Amer. J. Hum. Genet.* **44**, 388–396. 3) Zabel *et al.* (1989) *HGM* **10**, A 2407.

Dinucleotide repeat polymorphism at the human liver arginase gene (ARG1)

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Source/Description: The polymorphism is a dinucleotide repeat of the form (AC)17 and starts at position 643 of the human liver arginase gene on chromosome 6q23 (GenBank accession no. X12699) (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as previously described (2). The predicted size of the amplified sequence is 216 bp.

Primer Sequence:

AAGCCCCCATAACATAGAGTG (CA strand)
GCAGGACGTTTTTATGAATG (TG strand)

Frequency: Estimated from 112 chromosomes of unrelated CEPH individuals. Observed heterozygosity is 47% with a PIC of 0.43.

Allele (bp)	Frequency	Allele (bp)	Frequency
B1 222	0.223	B4 216	0.687
B2 220	0.009	B5 214	0.018
B3 218	0.063		

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

Chromosomal Localization: The human ARG1 gene has been assigned to chromosome 6q23 (3).

PCR Conditions: The PCR reaction was performed using 200 ng genomic DNA, 10 pMoles of each primer and 1.5 units of Taq polymerase in a final volume of 50 μ l. Samples were subjected to 30 cycles consisting of 40 seconds at 92°C, 30 seconds at 52°C and 30 seconds at 72°C.

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References: 1) Takiguchi, M. *et al.* (1988) *Nucleic Acids Res.* **16**, 8789–8802. 2) Weber, J.L. and May, P.E. (1989) *Amer. J. Hum. Genet.* **44**, 388–396. 3) Sparkes, *et al.*, (1986) *Am. J. Hum. Genet.* **39**, 186–193.