An *Eco*RI RFLP downstream of the human *c-myc* gene

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Source/Description: 1A4F (D8S197) is a 3.8 kb *Eco*RI genomic fragment mapped more than 100 kb downstream of the *c-myc* gene, and subcloned into pUC119. This probe is derived from cosmid clone #1A which was obtained by screening a cosmid library of human leukocytes (1) with a DNA fragment of the *c-myc* gene amplicon in GL-16 human glioblastoma cell line. Although we have not precisely determined yet the location of 1A4F, we suggest that its position is between 100 to 300 kb downstream of the *c-myc* gene, since 1A4F is within the amplicon of COLO320 but outside of the amplicon of HL60 (Lin and Shibuya, unpublished results) (2, 3).

Polymorphism: *Eco*RI identifies a two-allele polymorphism, 5.8 kb (A_1) and/or 3.8 kb (A_2).

Frequency: 50 unrelated Japanese individuals were studied.

A_1 : 0.18

A_2 : 0.82

Not Polymorphic For: *Hind*III, *Pst*I, *Kpn*I, *Sac*I, *Bam*HI.

Chromosomal Localization: 8q24.

Mendelian Inheritance: Co-dominant inheritance was observed in three two-generation families (a total of 17 individuals).

Probe Availability: Contact M. Shibuya.

Other Comments: This probe needs to be competed with sheared human placenta DNA because it contains human genomic repeated sequences (4). We are currently isolating a unique sequence from 1A4F clone. (Comments 1 and 2 from 10/30/91 letter).

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Two novel sequence polymorphisms of the human protein C gene

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Source/Description: A 287 bp fragment (nt 3174–3460 as in ref. 1) amplified as described by Yamamoto *et al.* (2) using the primer D-2' (nt 3431–3460) and the mutagenic primer M-1 (nt 3174–3203, with only one substitution of ³²⁰¹C→A) for PC-457 detection, and a 134 bp fragment (nt 6078–6211 as in ref. 1) amplified using the primer E-1 (nt 6078–6107) and the mutagenic primer M-2 (nt 6182–6211, with only one substitution of ⁶¹⁸⁴A→T) for PC-664 detection.

Polymorphism: A C→T transversion in the triplet coding for ⁸⁷Arg (CGC:F1→CGT:F2), named as PC-457 according to the nucleotide number of the protein C cDNA, and an A→G transition in the triplet coding for ¹⁵⁶Lys (AAA:G1→AAG:G2), named as PC-664 as described above, were found. MaeII (A/GCT) identifies the two sequence polymorphisms when used to digest the 287 bp fragment (F1) (to a 259 bp and a 28 bp fragment (F2)) and the 134 bp fragment (G1) (to a 104 bp and a 30 bp fragment (G2)).

Frequency: Studied in 28 unrelated Japanese individuals

PC-457: F1 = 0.875

PC-664: G1 = 0.643

F2 = 0.125

G2 = 0.357

heterozygosity rate =

heterozygosity rate

21.9%

= 45.9%

Chromosomal Localization: The protein C gene has been localized on chromosome 2 (3).

Mendelian Inheritance: Co-dominant segregation shown in 2 families, 12 individuals.

Primer Availability: Available for collaborative studies; write to Koji Yamamoto.

References: 1) Foster, D.C. *et al.* (1985) *Proc. Natl. Acad. Sci. USA* **82**, 4673–4677. 2) Yamamoto, K. *et al.* (1991) *Blood* **77**, 2633–2636. 3) Rocchi, M. *et al.* (1986) *Hum. Genet.* **74**, 30–33.

PC-457

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