A PstI polymorphism in the human coagulation factor V (F5) gene

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Source/Description: 1.5 kb cDNA cloned into Hinc II site of pUC13 (Wang *et al.*, 1988).

Polymorphism: Pst I identifies a two allele polymorphism with fragments of 10.8 kb and 8.5 kb. Constant bands at 2.66 kb, 2.31 kb, 2.26 kb and 1.66 kb are observed.

Frequency: Studied in 38 unrelated Caucasians:
10.8 kb allele (A1): 0.74 8.5 kb allele (A2): 0.26.

Not Polymorphic For: BamHI, EcoRI, HindIII, PvuII, TaqI with DNA from the same 38 unrelated individuals.

Chromosomal Localization: The F5 gene maps to 1q21–q25 (Wang *et al.*, 1988).

Mendelian Inheritance: Co-dominant segregation shown in 9 informative families with 43 children.

Probe Availability: Freely available from RTAM for research purposes.

Other Comments: Probe works well under normal stringency conditions.

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Reference: Wang, H., Riddell, D.C., Guinto, E.R., MacGillivray, R.T.A. and Hamerton, J.L. (1988) *Genomics* 2, 324–328.

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Three RFLPs at the 3' end of the cystatin C gene, the disease gene in hereditary cystatin C amyloid angiopathy (HCCAA) in Iceland

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Source and Description of Clone: C6a, a full length human cystatin C cDNA clone subcloned into the EcoRI site of pUC 18 (1). C3E2-B, a 1.2 kb HindIII/BamHI fragment of a genomic library clone, C3E2 (2).

Polymorphism: a) A polymorphic PstI site, 1.4 kb from the 3' end of the gene, hybridized with probe C6a reveals a two allele polymorphism with band sizes of 1.9 kb and 2.6 kb. b) A polymorphic SacI site, 4.1 kb from the 3' end of the gene, hybridized with probe C3E2-B reveals a two allele polymorphism with band sizes of 2.7 kb and 8.6 kb. c) A polymorphic EcoRI site, 16 kb from the 3' end of the gene, hybridized with probe C6a reveals a two allele polymorphism with band sizes of 18 kb and 21 kb.

Frequency: Results from 79 unrelated individuals:

PstI: 1.9 kb allele: 0.85; 2.6 kb allele: 0.15

SacI: 2.7 kb allele: 0.73; 8.6 kb allele: 0.27

EcoRI: 21 kb allele: 0.71; 18 kb allele: 0.29

Not Polymorphic For: AluI, AvaII, BamHI, DraI, EcoRV, HincII, HindIII, KpnI, MspI, PvuII, ScaI, StuI, TaqI, Tth111I, XbaI, and XhoI screened in 10 unrelated individuals.

Chromosomal Localization: Chromosome 20, based on human-rodent cell hybrids (3).

Mendelian Inheritance: A co-dominant segregation pattern is seen in 9 families with HCCAA.

Probe Availability: From M. Abrahamson.

Comments: A mutation (revealed as an additional 630 bp AluI fragment in patients) in the cystatin C gene causes hereditary cystatin C amyloid angiopathy (HCCAA), a fatal dominantly inherited disease in young adults in Iceland (4). The HCCAA mutation is only found in association with the haplotype determined by the EcoRI 21 kb, PstI 1.9 kb and SacI 2.7 kb alleles. Mild linkage disequilibrium is observed between the SacI and EcoRI alleles.

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References: 1) Abrahamson, M. *et al.* (1987) *FEBS Lett.* 216, 229–233. 2) Abrahamson, M. *et al.* (1990) *Biochem. J.* 268, 287–294. 3) Abrahamson, M. *et al.* (1989) *Hum. Genet.* 82, 223–226. 4) Palsdottir, A. *et al.* (1988) *Lancet* ii, 603–604.

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