Polymorphism detected by multiple RENS in the human coagulation factor II (F2) gene

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Source and Description: Clone pIIH13 has a 2.0 kb insert of F2 cDNA.

Polymorphism: PstI identifies alleles of 8.3 kb (A1) and 9.1 kb (A2) with constant bands of 1.8, 1.4 and 1.2 kb. The same polymorphism is detected by HindIII (B1: 6.9, B2: 7.5, bands at 5.5, 4.6 and 1.2 kb constant), SstI (C1: 5.9, C2: 6.7, constant 3.0 kb), TaqI (D1: 5.3, D2: 6.0, constant bands at 3.3, 1.7, 1.3 and 1.2 kb), BamHI (E1: 12.8, E2: 14.1, constant bands at 24, 9.6 and 2.7 kb).

Frequency: In 20 unrelated Caucasians PstI: 9.1 kb allele (A1): 0.70, 8.3 kb allele (A2): 0.30.

Not Polymorphic For: EcoRI and PvuII with DNA from the above unrelated individuals.

Chromosomal Localisation: F2 maps to 11p11-q12 (Royle et al., 1987).

Mendelian Inheritance: Codominant segregation shown in 8 informative families with 21 children.

Probe Availability: Freely available from R.T.A. MacGillivray for research purposes.

Other Comments: The RFLP variation does not correspond to the allelic variants of the protein detected after electrophoresis of plasma in agarose (Board, 1982).

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Isolation and mapping of a polymorphic DNA sequence (R7) on chromosome 12 (D12S52)

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Source and Description of the Clone: A 1 kb BglIII-BamHI genomic fragment subcloned into plasmid pUC 18. This fragment maps 12 kb centromeric of a previously described polymorphic DNA sequence (D12S33) (1). This latter had been assigned to chromosome 12q22-3.

Polymorphism: BglIII identifies a simple two allele polymorphism at either 5.5 or 3.6 kb, without constant band.

Frequency: Estimated from 32 unrelated Caucasians; A1: 5.5 kb 0.54 A2: 3.6 kb 0.46

Mendelian Inheritance: Co-dominant segregation of the BglIII RFLP is observed in 5 families (15 informative meioses).

Probe Availability: Will be available only for DNA polymorphism studies.

Other Comments: RFLP was observed under normal hybridization and wash stringencies.

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Figure 1: Lane 1: HindIII digested λ; Lane 2: A1; Lane 3: A2; Lane 4-6: A1A2.