MspI RFLP in intron 8 of the human protein C gene

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SOURCE/DESCRIPTION: a 297 bp fragment (nt 7728-8025 as in ref. 1) amplified as described by Saiki et al (2) using the primers 88-327 (nt 7728-7747 as in ref. 1) and 88-328 (nt 8006-8025 as in ref. 1). This fragment is located in intron 8 of the protein C gene.

POLYMORPHISM: MspI (C/CGG) identifies a two allele polymorphism when used to digest the 297 bp fragment (A1). Most commonly a MspI site is present at position 7932 and a 204 bp (A1) and a 93 bp fragment is produced. In some alleles (1%) the MspI site is absent (A2).

FREQUENCY: Studied in 128 unrelated European caucasians.

MspI: A1 = 0.99
A2 = 0.01

CHROMOSOMAL LOCALISATION: The protein C gene has been localized on chromosome 2 (3).

MENDELIAN INHERITANCE: Co-dominant segregation shown in 2 families, 15 individuals.

PRIMER AVAILABILITY: Available for collaborative studies; write to Pieter Reitsma.

OTHER COMMENTS: Overnight digestion is necessary to assure complete digestion.

REFERENCES:

1. Donald C. Foster et al (1985) Proc Natl Acad Sci USA **82**: 4673-4677
2. Saiki, RK et al (1988) Science **239**: 487-491
3. Rocchi et al (1986) Hum Genet **74**: 30-33

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