A CCA/CCG neutral dimorphism in the codon for Pro 626 of the human protein S gene PSα (PROS1)

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Exon 15 of the human protein S gene PSα (PROS1) was amplified using the polymerase chain reaction. Sequence analysis showed an A/G neutral dimorphism at the third position of the codon for Pro^{626} of the PSα gene. The dimorphism can be screened with allele specific oligonucleotides (see figure).

Primer Sequences: PS 15 A (CAAGATGCTAAAAGTCTTGG) and PS 15 B (GATAGCAAGAGAAGTAAGAATTTC) were used in the PCR. PS 626 A (CTCATGTCCATCAGTTTGG) and PS 626 G (CTCATGTCCGTCAGTTTGG) were used as allele specific oligonucleotides in the hybridization.

PCR conditions: Amplifications, 5′-end labelling of PS 626 A and PS 626 G and the oligonucleotide hybridization of the PCR fragments were performed essentially as described (2).

Chromosomal Localization: The human protein S locus that consists of the active PSα gene and the pseudogene PSβ is localized on chromosome 3p11.1→3q11.2 (3, 4).

Frequency:
CCG allele (A1) 0.48
CCA allele (A2) 0.52
studied in 28 unrelated European Caucasians.

Mendelian Inheritance: Demonstrated in two families.

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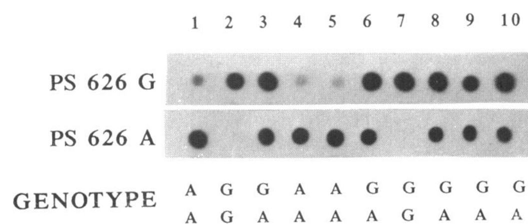

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A *Mae*III polymorphism within intron A of the insulin (INS) gene detectable by PCR

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Source/Description: Oligonucleotide primers were used to amplify a 350 bp genomic fragment from within intron A of the insulin gene. A sequencing variant from within this region had previously been described (1).

PCR Primers:
5′GCTGCATCAGAAGAGGCCATC 3′
5′ACACTAGGTAGAGAGCTTCCA 3′

Method: 1 μg of genomic DNA was amplified using 0.5 μg of each primer in a total volume of 100 μl containing 1×*Taq* polymerase buffer (BCL) and 200 μM dNTPs. Samples were heated to 97°C for 5 min after which 2 units of *Taq* polymerase (BCL) were added. Reactions were cycled 30 times at 94°C for 1 min, 58°C for 1 min and 74°C for 1 min. Amplified products and restriction enzyme digests (10 μl of amplified product in a total volume of 50 μl) were analysed by electrophoresis through a 3% agarose gel.

Polymorphism: Digestion with *Mae*III reveals a two allele system, allele R1 is 350 bp and allele R2 is 197 bp and 153 bp.

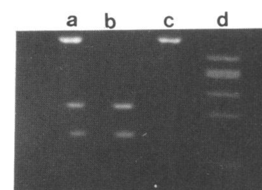
Frequency: Determined from a population of 50 unrelated individuals
Allele R1: 0.37
Allele R2: 0.63
The frequency of heterozygotes is 42% (21 out of 50 individuals).

Mendelian Inheritance: Codominant segregation was observed in three families (14 individuals).

Chromosomal Localisation: The insulin gene has been localised to 11p15.5 (2).

Acknowledgements: This work was supported by RMCH from the Pendlebury leukaemia Research fund.

References: 1) Ulrich,A. *et al.* (1980) *Science* **209**, 612–615. 2) Bell,G.I. *et al.* (1982) *Nature* **295**, 31–35.



The figure shows amplification products from three individuals digested with *Mae*III (a–c). Size marker (d) is ΦX174 *Hae*III digest.

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