HinfI polymorphism in the human progesterone associated endometrial protein (PAEP) gene

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Source and Description: pPP14-1 probe is a 900 bp cDNA corresponding to the coding region of PAEP gene cloned into EcoRI site of pGEM-3 blue vector (1).

Polymorphism: HinfI identifies a two allelic polymorphism with bands 1000 bp (allele A1) or 600 bp and 400 bp (allele A2).

Frequency: Studied in 56 unrelated Finnish individuals:

allele A1 (1000 bp) 0.05

allele A2 (600 bp and 400 bp) 0.95

Not Polymorphic For: BamHI, BglII, EcoRI, HincII, HindIII, PstI, SacI, XbaI (tested on 21 unrelated individuals).

Chromosomal Location: PAEP gene has been localized to 9q34 (2).

Probe Availability: Contact Dr Mervi Julkunen (1).

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References: 1) Julkunen *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 8845–8849. 2) Van Cong *et al.* (1991) *Hum. Genet.* **86**, 515–518.

Figure 1. HinfI-digested human DNA samples hybridized with pPAEP cDNA. Lanes 1 and 10 genotype A1A2, lanes 2–9 genotype A2A2.

DdeI polymorphism within sequence encoding 3' untranslated region of c-Ki-ras2 (KRAS2)

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Source/Description: PCR primers for the amplification of a 375 bp region of DNA encoding part of c-Ki-ras exon 4b and immediate downstream sequence (1). Primer 961 is within the exon and 962 within the region encoding the 3' untranslated sequence.

PCR Primers:

961 5' GAAAAAGAAGTCAAAGACAAAGTGTG 3'

962 5' GCTTCATTAATTTGTTTCACACCAAC 3'

Polymorphism: DdeI identifies a simple two allele polymorphism. Constant band 95 bp.

J1 280 bp

J2 210 and 70 bp.

Frequency: Studied in 65 unrelated individuals

J1 0.52

J2 0.48

Chromosomal Localisation: c-Ki-ras2 has been localised to 12p12.1.

Mendelian Inheritance: Codominant segregation has been demonstrated in five families (25 individuals).

PCR Conditions: 1 µg of genomic DNA was amplified in a 100 µl reaction using 1 µg of oligo 962 and 0.25 µg of 961 in Taq reaction buffer (BCL) containing dNTPs (0.25 mM). After heating at 97°C for 4 min 2.5 units of Taq polymerase (BCL) was added and reactions were cycled 30 times at 94°C for 1 min, 62°C for 1 min and 74°C for 1 min.

Comment: The polymorphic site lies within a region encoded by the Ki-ras mRNA molecule (1). After PCR amplification of cDNA (in heterozygous individuals) the polymorphism can therefore be used to monitor allele specific expression by comparing relative band intensities after DdeI digestion. Any imbalance can be quantified by standard techniques.

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Reference: 1) McGrath *et al.* (1983) *Nature* **304**, 501–506.

Figure shows amplification products from five individuals digested with DdeI. Outside tracks are ΦX174 HaeIII digests.