A TaqI polymorphism in the human cyclin A gene

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Source/Description: Probe HHT2 is a 1.2 kb HindIII genomic fragment from the 3' part of the human cyclin A gene (1) cloned into the Bluescript plasmid.

Polymorphism: TaqI detects a two allele polymorphism
A1 = 1.5 kb
A2 = 4 kb

Frequency: Estimated from 31 unrelated individuals
A1 = 0.71
A2 = 0.29

Not Polymorphic For: PstI, BamHI, EcoRV.

Chromosomal Localization: Localized to 4q27 by in situ hybridization (2).

Mendelian Inheritance: Codominant segregation was observed in five 2 generation families and two 3 generation families.

Probe Availability: C. Bréchot, U75 C.H.U. Necker, 156 rue de Vaugirard, 75015 Paris. Tél: (1) 40.65.99.12; Fax: 45.67.53.33.

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PCR detection of the frequent TaqI RFLP at locus D21S13E

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Source/Description: We recently published oligo primer sequences for the PCR detection of a high frequency EcoRI polymorphism and a low frequency TaqI polymorphism at the D21S13 locus (1). Here we report primer sequences defining a 389 bp fragment overlapping the frequent TaqI polymorphic site (2).

PCR Primers:
PSCR13-K: 5' - GGG TTC TAA AGG GAA GAA AG- 3'
PSCR13-L: 5' - CCT AAC AGA GGT CAC AAG GA- 3'

Polymorphisms: After a TaqI digest of the amplified fragment two alleles can be identified: A1: 389 bp, A2: 313 bp + 76 bp.

Frequency:
A1: 0.60 A2: 0.40 (3).

Chromosomal Location: pGSM21 (D21S13) is located in the 21q11.2 region (4).

Mendelian Inheritance: Codominant inheritance of this polymorphism was demonstrated in two extended families with Alzheimer's disease (106 individuals).

PCR Conditions: The PCR reaction is carried out in a total volume of 50 µl containing approximately 250 ng DNA, 2 units Taq DNA polymerase, 50 pmol of each primer, 200 µM dNTP's, 10 mM Tris-HCl pH 8.3, 50 mM KCl, 1.5 mM MgCl2, 0.001% gelatin. The amplification is performed for 30 cycles with an annealing temperature of 60°C. The amplified product is digested with TaqI and the DNA fragments are analyzed on 1.5% agarose gels.