An StuI RFLP at the human cholesteryl ester transfer protein (CETP) locusDilys Freeman¹, James Shepherd, Christopher J. Packard, Stephen E. Humphries² and Dairena Gaffney

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SOURCE/DESCRIPTION: 1581 base pair CETP cDNA clone as described (1) or a 732bp PstI-PvuII (732P) subfragment of this (2).

POLYMORPHISM: Digestion with StuI and probing with 732P probe detects two allelic fragments of 4.0 and 4.3 kb.

FREQUENCY: Studied in 65 unrelated individuals.

S1 allele 4.0 kb frequency 0.92

S2 allele 4.3 kb frequency 0.08

Invariant bands with CETP 1581 probe at 5.8 kb, 4.8 kb and 3.4 kb.

S1/S2 alleles difficult to detect with this probe. 732P probe shows the same invariant band at 3.4 kb.

NOT POLYMORPHIC FOR: AluI, ApaI, BamHI, BanII, BglI, BglII, BstEII, ClaI, DraI, EcoRI, EcoRV, HaeIII, HincII, HindIII, HinfI, HhaI, HpaI, KpnI, PstI, PvuII, RsaI, SalI, SmaI, SstI, XbaI, XhoI.

CHROMOSOMAL LOCATION: 16q21 (3)

MENDELIAN INHERITANCE: Demonstrated in one family (see Figure).

PROBE AVAILABILITY: Contact Dennis Drayna, Genentech, 460 Pt. San Bruno Boulevard, South San Francisco, CA 94080, USA.

OTHER COMMENTS: This polymorphism was first detected using the total CETP cDNA probe as presence or absence of the 4.0 kb band. In the presence of 5.8 and 4.8 kb bands the total amount of radioactivity that bound to the band at 4.0 kb and its allele, the 4.3 kb band, was sufficiently low to make detection of these bands difficult. Hence the use of the 732P probe. For 45 individuals the TaqI RFLPs (1) in the CETP gene were also determined and these appear to be in linkage equilibrium with the StuI RFLP.

REFERENCES:

(1) Drayna D. and Lawn R. (1987). NAR 15, 4698.

(2) Drayna et al (1987). Nature 327, 632-634.

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FIGURE: Mendelian codominant inheritance of the StuI RFLP shown. The first six tracks have been probed using the 1581 bp probe and the last six probed using the 732P probe. A = Mother, B = Father, C = Son, D = Daughter, E, F, G = Unrelated Individuals.