AccI polymorphism in von Willebrand factor (F8VWF) at codon 516

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Source/Description: The sequence of VWF (1) shows a potential dimorphism of the third base of codon 516. An AccI site is (+) when the sequence is TAC and (−) when it is TAT.

Polymorphism: Using two 17 base primers starting 38 bp 5' and 118 bp 3' to the dimorphic site, genomic DNA was amplified 35 cycles as described (2) under these conditions: 30'' at 92°, 4' at 60°, electrophoresis in 12% PAG. AccI (−, −) shows one 157 bp band (S1); (+, +) shows two (123, 34 bp) (S2) (+, −) shows all three (Fig. 1).

Primers: Primer 1: 5' AGA GTG GCC TGG TCT CT 3'

Primer 2: 5' TGC AGC TTC CAG GCG TT 3'

Chromosomal Location: 12pter-p12. Codon 516 of VWF.

Mendelian Inheritance: Autosomal co-dominant in one VWD family (Fig. 2).

Allelic Frequencies: The (+:−) values were: Anglo-Americans (64 chromosomes) .24:.76; Swedes (60) .35:.65; Basques (90) .36:.64; East Indians (54) .26:.74; Chinese (50) .22:.78; Malays (52) .23:.77; African-Americans (54) .56:.44; East Africans (50) .60:.40.

Heterozygosity (Expected/Observed): Anglo-American (.37/.42); Swedes (.46/.63); Basques (.46/.49); East Indians (.39/.46); Chinese (.34/.44); Malays (.35/.31); African-Americans (.49/.67); East Africans (.48/.40).

Allelic Association: AccI (−) is strongly associated with RsaI (+) in 267 non-African chr. (3): $D = .099$, $r = .487$, $P < .01$. In 104 African chr. AccI (+) is strongly associated with RsaI (−): $D = .08$, $r = .32$, $P < .01$. The distance between the AccI and RsaI RFLP is estimated to be approximately 12.8 kb.

References: 1) Mancuso *et al.* (1989) *JBC* **264**, 19514–19527. 2) Graham *et al.* (1989) *Blood* **73**, 2104–2107. 3) Kunkel *et al.* (1990) *Nucl. Acids Res.* **18**, 4961.

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