Two additional TaqI RFLPs in von Willebrand factor gene (VWF) and pseudogene

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SOURCE/DESCRIPTION: A 4 Kb cDNA fragment coding for the N- terminal portion of the mature vWF and inserted in the EcoRI site of pBR322.

POLYMORPHISMS: Two TaqI polymorphisms with alleles of 1.0 (A₁) and 2.2Kb (A₂) and of 0.7 (P₁) and 1.8 (P₂) respectively (see figure). Several constant bands are present.

FREQUENCIES: Estimated in 25 unrelated Italian subjects; 0.78 (A₁) and 0.22 (A₂); 0.86 (P₁) and 0.14 (P₂).

NOT POLYMORPHIC FOR: BglII and BamHI.

CHROMOSOMAL LOCALIZATION: A₁A₂, 12p12-12pter; P₁P₂, pseudogene on chromosome 22 (Shelton-Inloes et al. 1987), shown in a subject with complete deletion of vWF gene.

MENDELIAN INHERITANCE: Codominant segregation shown in four families.

PROBE AVAILABILITY: Available without restriction. Write to Professor D. C. Lynch.

OTHER COMMENTS: Detectable at wash stringency 2XSSC 60° C.

REFERENCE: Lynch et al. (1985) Cell 41, 49. Bernardi et al. (1987) N.A.R. 15, 1347. Shelton-Inloes et al. (1987) J. Clin. Invest. 79, 1459.

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