RsaI RFLP in the human von Willebrand factor gene

M.C. Iannuzzi, B.A. Konkle, D. Ginsburg and F.S. Collins

University of Michigan Medical Center and Howard Hughes Medical Institute, Ann Arbor, MI 48109, USA

SOURCE/DESCRIPTION: pVWE6, a 2 kb Eco RI/SacI fragment of the von Willebrand factor cDNA inserted in pUC13 (Ginsburg et al., 1985). The fragment used as probe was a 232 bp segment obtained from a PstI digestion of pVWEB.

POLYMORPHISM: RsaI (GT/AC) (New England Biolabs) identifies an invariant band at 580bp and a simple two allele polymorphism with band at either 1.0kb (A1) or 660bp (A2).

FREQUENCY: Estimated in 20 unrelated Caucasians,
1.0kb allele (A1) 0.22
660bp allele (A2) 0.78

NOT POLYMORPHIC FOR: PVWEB identifies BglII (Verweij et al., 1985) and TaqI (Bernardi et al., 1987).

CHROMOSOMAL LOCALISATION: The probe has been localized to the region 12p12-12pter by in situ hybridization (Ginsburg et al., 1985).

MENDELIAN INHERITANCE: Co-dominant segregation demonstrated in a family of 19 individuals.

PROBE AVAILABILITY: Probe freely available.

OTHER COMMENTS: No problem on RFLP analysis under normally stringent conditions.