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A polymorphic Xba I site within the human von Willebrand factor (vWF) gene identified by a vWF cDNA clone

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SOURCE/DESCRIPTION A 770 bp. Pst I fragment, derived from the human vWF cDNA clone pvWF1210 (Verweij et al. 1985), subcloned in pUC18.

POLYMORPHISM Xba I (T/CTAGA) identifies two invariant bands at 3.8 kb and 2.8 kb and a simple two-allele polymorphism with a band at either 6.9 kb or 5.2 kb.

FREQUENCY Studied 20 European Caucasians.
6.9 kb allele (A_1) : 0.87
5.2 kb allele (A_2) : 0.13

NOT POLYMORPHIC FOR Ava I, Bst I, Eco RI, Eco RV, Hae III, Hind III, Msp I, Pst I and Pvu II.
All with a panel of 10 unrelated Caucasians.

CHROMOSOMAL LOCALISATION Probe localized to 12p12-12pter by *in situ* hybridization (Ginsburg et al. 1985)

MENDELIAN INHERITANCE Co-dominant segregation demonstrated in two families, 46 individuals.

PROBE AVAILABILITY Probe freely available for diagnostic screening (write to H.P.).

OTHER COMMENTS No problems on RFLP analysis under normally stringent conditions.

REFERENCE Verweij et al., Nucleic Acids Research 13 (1985) 4699-4717.
Ginsburg et al., Science 228 (1985) 1401-1406.

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