An EcoRI polymorphism in the human von Willebrand factor (vWF) gene

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SOURCE/DESCRIPTION: A 3.6 kb Xmn I-Sal I fragment of the von Willebrand factor cDNA inserted in pGEM 2.

POLYMORPHISM: Eco RI (G/AATTC) identifies several invariant bands and a single two allele polymorphism with either one band at 7.0 kb (A 1) or two bands at 5.3 kb and 1.7 kb (A 2).

FREQUENCY: Studied in 101 European Caucasians.

7.0 kb allele (A 1): 0.865
5.3 kb and 1.7 kb allele (A 2): 0.135

POLYMORPHIC FOR: Bgl II (Verweij et al. 1985), Bam HI (Nishino and Lynch 1986), Xba I (Quadt et al. 1986; Lavergne et al. 1987), Sac I (Konkle et al. 1987).

CHROMOSOMAL LOCALISATION: 12p12-12pter (Ginsburg et al. 1985).

MENDELIAN INHERITANCE: Co-dominant segregation shown in 5 families.

PROBE AVAILABILITY: Probe available without restriction. Write to D. C. Lynch.

OTHER COMMENTS: No problems on RFLP analysis under normally stringent conditions. This RFLP can also be detected with the 1.9 kb Sac I-Sac I fragment of the von Willebrand factor cDNA.


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