StuI polymorphisms in the vWF gene

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Source and Description of Clone: The 368 bp BamHI/SacI fragment of pvWH22 was used as a probe. pvWH22 is a 4 kb partial cDNA clone for human vWF inserted into pUC 13 (Ginsburg *et al.*, 1985).

Polymorphisms: Two StuI (AGGCCT) (New England Biolabs) polymorphisms identify two invariant bands of 8.4 kb and 7.0 kb and four variant bands with alleles at 5.6 kb (A1) and 4.8 kb (A2) and 3.8 kb (B1) and 3.3 kb (B2) (see figure).

Frequency: 23 unrelated caucasian individuals.
5.6 kb (A1) – 0.3
4.8 kb (A2) – 0.7
3.8 kb (B1) – 0.6
3.3 kb (B2) – 0.4

There was no evidence for linkage disequilibrium between the two polymorphisms.

Not Polymorphic For: EcoRI, BamHI, SacI, in a panel of 10 unrelated individuals.

Chromosomal Localization: 12p12–12pter (Ginsburg *et al.*, 1985). The 3' region of BamHI/SacI fragment corresponds to the 5' boundary of an area that could possibly hybridize with similar sequences on human chromosome 22 (Sadler *et al.*, 1987). The region of vWF cDNA contained in the BamHI/SacI fragment corresponds exclusively to sequences on chromosome 12 (Inbal *et al.*, 1989).

Mendelian Inheritance: Co-dominant segregation demonstrated in one family of 11 individuals.

Probe Availability: Available from Robert I.Handin.

Other Comments: No problems on RFLP analysis under normally stringent conditions.

Acknowledgement: This work was supported by NIH P01 HL33014.

References: Ginsburg *et al.* (1985) *Science* **228**, 1405. 2) Sadler *et al.* (1987) *J. Clin. Invest.* **79**, 1459. 3) Inbal *et al.* (1989) *Nucl. Acids Res.* **17**, 10143.

Two polymorphisms at the D10S94 locus

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Source/Description: A 0.9 kb DNA sequence isolated by Alu element-mediated PCR from the somatic cell hybrid H2CL1 (Brooks-Wilson *et al.*, 1990) and cloned into the SalI site of pUC18.

Polymorphism: 2 allele codominant polymorphisms identified with PvuII (A1 = 2.5 kb, A2 = 1.8 kb) and RsaI (B1 = 8.0 kb, B2 = 7.0 kb).

Allele Frequencies:

A1 0.67
A2 0.33 94 chromosomes
B1 0.97
B2 0.03 38 chromosomes

Not Polymorphic For: TaqI, MspI, BglII, SacI, HindIII and EcoRI.

Mendelian Inheritance: Codominant inheritance observed in several *MEN2A* kindreds.

Chromosomal Localization: Localized to 10 cen-q11.2 using somatic cell hybrids. Linkage with markers from the pericentromeric region confirms the localization.

Probe Availability: Contact P.J.Goodfellow.

Other Comments: Slight background smear with normal hybridization/washing conditions. Higher stringency conditions required.

Reference: Brooks-Wilson, A.R., Goodfellow, P.N., Povey, S., Nevanlinna, H.A., de Jong, P.J. and Goodfellow, P.J. *Rapid cloning and characterization of new DNA markers from the pericentromeric region of chromosome 10 by Alu element-mediated PCR.* submitted.