Dinucleotide repeat polymorphism at the human thrombospondin gene (THBS1)

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Source/Description: The polymorphic (CT)_n repeat begins at base pair 2595 in intron A of the human thrombospondin gene on chromosome 15q15 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 165 bp.

Primer Sequences: CTGATCTTGCTCACCTTCGA (CT strand); GCGTTTGCTGAAATGAAGGA (GA strand).

Frequency: Estimated from 50 chromosomes of unrelated individuals. Heterozygosity = 56%.

Allele (bp)	Frequency	Allele (bp)	Frequency
A1 181	0.02	A6 171	0.02
A2 179	0.02	A7 169	0.14
A3 177	0.02	A8 167	0.06
A4 175	0.02	A9 165	0.60
A5 173	0.10		

Mendelian Inheritance: Co-dominant segregation was observed in two informative families.

Chromosomal Localization: THBS1 gene has been assigned to chromosome 15q15 (3).

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described (4) except that the denaturation cycle at 94°C was extended to 1.4 minutes. The dinucleotide repeat was based on a (CT)₁₄ sequence.

References: 1) Donoviel, D.B. *et al.* (1988) *J. Biol. Chem.* **263**, 18590–18593. 2) Weber, J.L. and May, P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Jaffe, E. *et al.* (1990) *Genomics* **7**, 123–126. 4) Weber, J.L. *et al.* (1990) *Nucl. Acids Res.* **18**, 4637.

An MspI polymorphism in the von Willebrand Factor gene

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Source/Description: Two primers (W17 and W18) were used to amplify a 226 bp long fragment including exon 19 and a part of introns 18 and 19 of the human von Willebrand Factor gene (VWF) (1).

Primers:

W17 = AGGGCTCTAGATCAGTCACTGTGGCCCT

W18 = TGGCCGCGAGCTCCCTCACTCCACC

Polymorphism: After PCR amplification (30 cycles: 1 min at 94°C, 1 min at 55°C, 2 min at 72°C), the amplified fragments were digested by MspI restriction endonuclease. Two MspI sites were present in the published sequence (1) but one of these sites, 23 bp in 3' of exon 19, was shown to be either present (allele +: 125 + 59 + 42 bp) or absent (allele -: 167 + 59 bp).

Frequency: Estimated in 50 unrelated Caucasians
allele +: 0.33 allele -: 0.67

Chromosomal Localization: 12p12–12pter (2).

Mendelian Inheritance: Co-dominant segregation shown in 2 families.

References: 1) Mancuso, D.J. *et al.* (1989) *J. Biol. Chem.* **264**, 19514–19527. 2) Ginsburg, D. *et al.* (1985) *Science* **228**, 1401–1426.

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