An *MspI* polymorphism at the DXS11 locus

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Source and Description of Clone: The probe, QOG13.5E, is a 1.2 kb fragment cloned into the *EcoRI* site of pUC 19. The fragment was subcloned from an EMBL3 phage clone isolated from a total human library after screening with p22.33 (1).

Polymorphism: QOG13.5E detects a simple two allele RFLP, B1 = 5.1 kb and B2 = 2.8 kb, with *MspI*. A single faint constant band was observed at 6.5 kb.

Frequency: In 9 unrelated females. B1 = 0.22, B2 = 0.78. Heterozygosity = 0.44.

Other Enzymes Tested: Negative in 9 unrelated females. *AluI*, *BamHI*, *BglI*, *BglII*, *DraI*, *EcoRI*, *EcoRV*, *HincII*, *HindIII*, *PstI*, *TaqI*, *XbaI*.

Chromosomal Localization: QOG13.5E maps within 16 kb of p22.33. p22.33 (DXS11) has been localized to Xq24-q25.

Mendelian Inheritance: The *MspI* RFLP has been found to show co-dominant Mendelian inheritance in a three generation family.

Probe Availability: Freely available for research purposes.

Other Comments: Haplotype analysis of the 9 females tested for the *MspI* RFLP detected by QOG13.5E and the *TaqI* RFLP detected by p22.33 gave observed heterozygosities of 0.44 and 0.33 respectively and a combined observed heterozygosity of 0.67.

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Reference: 1) Kidd *et al.* (1989) *Cytogenet. Cell. Genet.* **51**, 622–947.

Acyl-RFLP in intron 8 of hTPO gene

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Source/Description: We used two primers (TPO-8a and TPO-8b) to amplify a 755bp long fragment of the TPO gene including exon 8 plus 18bp upstream and 219bp downstream of this exon.

Primers: TPO-8a 5'-ACTCCCCTTTGCCTGCAGCT-3'
TPO-8b 5'-CTGCACAGGAGCTCAGATG-3'

Polymorphism: After PCR amplification (1) the products were digested by *Acyl* restriction endonuclease and electrophoresed on 6% Nu Sieve Agarose. The published sequence (2) reports five *Acyl* sites on this target. The two bands due to the *Acyl* site located 114bp downstream of exon 8 resulted clearly polymorphic: the 64bp and 59bp long fragments (G1) were replaced in some examples by one band of 123bp (G2) (loss of *Acyl* site). The other expected bands were present in all the samples (Fig. 1).

Frequency: Frequencies estimated in 49 unrelated Caucasians
G1 Allele +: 0.46 G2 Allele -: 0.54

Chromosomal Localization: 2pter-p12 (3).

Mendelian Inheritance: Co-dominant segregation shown in one pedigree (18 individuals with 8 significant meioses).

Comments: Inclusion of 9% DMSO (4) was necessary for generating the specific 755bp product. Not polymorphic for *HinfII*, *PstI*, *AvaII*, *MspI*, *TaqI* in the same sample group.

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References: 1) Rose *et al.* (1991) *Nucl. Ac. Res.* **19**, 4021. 2) Kimura *et al.* (1987) *Proc. Natl. Acad. Sci. USA* **84**, 5555–5559. 3) De Vijlder *et al.* (1988) *Cytogen. Cell Genet.* **47**, 170–172. 4) Smith *et al.* (1990) *Amplifications* **5**, 16–17

Acyl RFLP of the hTPO amplified region (intron 8) a: -/-; b: +/+.

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