

Two RFLPs at the TNP1 locus

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Source/Description: The Human Transition Protein 1 gene (TNP1) probe (1) used was a 700 bp genomic DNA sequence cloned into the SmaI site of pUC8 and was supplied by W.Engel.

Polymorphisms: TNP1 identifies a two allele polymorphism with TaqI: A1 = 9.6 kb; A2 = 8.4 kb.

Frequency: Analysis of 20 unrelated individuals yielded frequencies: A1 = 0.23; A2 = 0.77.

Not Polymorphic For: BanI, BclI, BglI, BstEII, EcoRI, HaeIII, HindIII, MspI, NcoI, PstI, PvuII, RsaI in a screen of 10–20 unrelated individuals.

Mendelian Inheritance: Autosomal co-dominant segregation has been demonstrated in 3 families.

Probe Availability: Contact Hannelore Kremling, Institut für Humangenetik der Universität, Gossler Strasse 12D, 3400 Göttingen, FRG.

Source/Description: Oligonucleotide primers from sequence information of the TNP1 gene were used to amplify a 1.7 kb region encompassing the gene.

PCR Primers:

5' CCA TTG ATG TTG ACA GTA GCA 3' (TN1)

5' CAC CTA GCT CAG GAA CTC AA 3' (TN2)

Polymorphisms: MspI identifies a two-allele polymorphism with bands of 1059 bp (B1), 355 + 704 bp (B2) and a constant band of 633 bp. PflMI identifies a similar polymorphism.

Frequencies: Analysis of 30 unrelated individuals yielded frequencies of: B1 = 0.45; B2 = 0.55. and heterozygote frequency = 0.60 for both MspI and PflMI.

Not Polymorphic For: BsaAI, BstNI, HhaI, StuI, TaqI in 10–20 unrelated individuals.

Chromosomal Localization: TNP1 has been localized to 2q35-q36 by somatic cell hybridization and *in situ* hybridization (1).

Mendelian Inheritance: Autosomal co-dominant segregation has been demonstrated in 9 families.

Other Comments: The MspI polymorphism is in linkage disequilibrium with the PflMI polymorphism.

PCR Conditions: 0.5–1 µg of genomic DNA was amplified with 5 pmoles of each primer, 1 unit Taq polymerase (Cetus), 200 µM dNTPs, in Cetus reaction buffer. PCR amplification was for 30 cycles at 94°C for 1.4 min, 58°C for 2 minutes.

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Reference: 1) Luerssen *et al.* (1990) *Genomics* **8**, 324–330.

Dinucleotide repeat polymorphism in the human coagulation factor XI gene, intron B (F11), detected using the polymerase chain reaction

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Source/Description: The polymorphic sequence (CA)₁₁ starts at base 212 of intron B of the human coagulation factor XI gene (1). The sequence was identified from a search of the EMBL and GenBank DNA sequence databases (GenBank M21185; M18297). The predicted amplified sequence length is 360 bp.

Primer Sequences:

5'TCACCCAAGTAGTGAACACAGC3'

5'GGTTGTTCCACCTGTAATCC3'

Frequency:

Allele	Size (bp)	Frequency
D1	376	0.47
D2	372	0.53

Heterozygosity 45% (estimated using 40 unrelated Caucasian individuals)

Mendelian Inheritance: Observed in CEPH families 1029 (Utah pedigree K1345) and 982 (Utah pedigree K1331).

Other Comments: 30 cycles of PCR were performed (2) using 200–300 ng of genomic DNA and 150 ng of each primer in a final reaction volume of 50 µl. The reaction mix contained a final magnesium concentration of 1.5 mM, 10% dimethyl sulphoxide (DMSO) and 0.1 mM trimethylammonium chloride (TMAC). Primers were designed using the program PRIMER (3).

Chromosomal Localization: 4q35.

References: 1) Asakai,R., Davie,E.W. and Chung,D.W. (1987) *Biochemistry* **26**, 7221–7228. 2) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Lowe,T., Sharefkin,J., Yang,S.Q. and Dieffenbach,C.W. (1990) *Nucl. Acids Res.* **18**, 1757–1761.