A TaqI polymorphism in the human NF1 gene

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Source and Description: A 193 bp cDNA fragment corresponding to exon 4 of the published cDNA sequence (1) for the human NF1 gene was amplified by PCR using 5'-ATAATTGTTG-ATGTGATTTTCATTG as forward primer and 5'-AATTTTG-AACCAGATGAAGAG as reverse primers. This cDNA was used as a probe for hybridization of southern blots made from human DNA samples.

Polymorphism: TaqI digestion yields two bands of 7.0 kb and 6.5 kb without constant band.

Frequency: Studied in a total of 40 unrelated Caucasians (20 males and 20 females)
B1 7.0 kb allele: 0.4
B2 6.5 kb allele: 0.6
Frequency of heterozygosity: 0.48.

Not Polymorphic For: EcoRI, PstI, PvuII in 10 unrelated Caucasians.

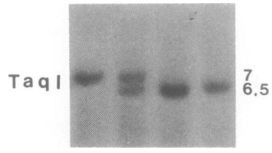
Chromosomal Localization: Assigned to 17q11 within NF1 gene (1).

Mendelian Inheritance: Co-dominant segregation of the TaqI RFLP was observed in two informative von Recklinghausen Neurofibromatosis (NF-1) families (10 meioses). Cosegregation with the NF-1 phenotype was observed in all these families.

Probe Availability: contact W.Xu.

Acknowledgement: This work was supported by LINK, the UK Neurofibromatosis Association.

Reference: 1) Cawthon,R.M. *et al.* (1990) *Cell* **62**, 193–201.



PCR detection of a repeat polymorphism within the F7 gene

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The human coagulation factor VII gene (F7) contains five regions of tandem repeats, four located in introns and one in an untranslated portion of the exon 8 (1). The presence of six or eight copies of a monomer repeat element has been reported in two clones of the FVII gene (2).

PCR amplification of FVII gene regions containing repeated and exonic sequences was performed and a polymorphism was found in the intron 7.

PCR Primers:
Sense oligo: 5'AATGTGACTTCCACACCTCC 3'
Antisense oligo: 5'GATGTCTGTCTGTCTGTGGA 3'

Polymorphism: The primers amplify two alleles A1 = 480 and A2 = 443 bp, which differ in one monomer element (37 bp).

Frequency: Estimated in 23 unrelated subjects
A1 = 0.30
A2 = 0.70

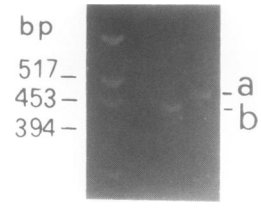
Chromosomal Localization: FVII gene has been localized to 13q34.

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

PCR Conditions: PCR amplifications are carried out in a volume of 25 μl, containing 0.1 μg human DNA, 7 pmoles of each primer, 200 μM dNTPs, 50 mM KCl, 10 mM Tris-Cl pH 8.3, 1.5 mM $MgCl_2$, 5% DMSO 1.6 units Taq polymerase. Cycles (30): 92°C for 20 sec, 57°C for 3 sec and 70°C for 40 sec. The products were electrophoresed on 2% agarose gel in 1×TAE buffer.

Acknowledgements: This work was supported by Ric San Final Reg Emilia Romagna and by P.F. CNR Ing Genet N 9100021PF99.

References: 1) O'Hara,P.J., Grant,F.J., Haldeman,B.A., Gray,C.L., Insley,M.Y., Hagen,F.S. and Murray,M.J. (1987) *Proc. Natl. Acad. Sci. USA* **84**, 5158–5162. 2) O'Hara,P.J. and Grant,F.J. (1988) *Gene* **66**, 147–158.



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