Dinucleotide repeat polymorphism at the D11S420 locus

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Source and Description of Clone: Cosmid 8.5 was from a human chromosome 11q-specific library (1). p8,5-3, a subclone of this cosmid, was sequenced and the sequences flanking a (GT)₁₈ repeat (EMBL accession number X52290) were used to design PCR primers.

PCR Primers:

(# 506)-5'-AGTTACACCGGTTCTGCAGA-3'

(# 507)-5'-GATTAATGATAGTGCTATCC-3'

Polymorphism: Allelic fragments were detected on DNA sequencing gels. Lengths (nt) were: A1=188, A2=190, A3=192, A4=194, A5=196, A6=208.

Frequencies: From 33 unrelated European Caucasians. A1=.39, A2=.36, A3=.063, A4=.063, A5=.063, A6=.063. The PIC calculated from these frequencies is 0.66.

Chromosomal Localization: 11q23.3-q24, by fluorescent in situ hybridization (1).

Mendelian Inheritance: Mendelian inheritance was observed in 2 informative CEPH families with a total of 22 children.

PCR Conditions: PCR was performed in a total volume of 25 μ l containing: 50 ng genomic DNA, 5 pmoles of each primer, 1.5 mM MgCl₂, 200 μ M dNTPs, 50 mM KCl, 10 mM Tris-Cl⁻, pH 8.3, 0.6 units Taq polymerase (Perkin-Elmer/Cetus) and 0.01% gelatin. Primer # 506 was 5' end labeled with ³²P. Amplification was for 30 cycles with denaturation at 94°C for 1 min, annealing at 49°C for 1 min and extension at 72°C for 30 sec. Amplified products were resolved on DNA sequencing gels and detected by autoradiography.

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Reference: 1) Lichter, P., Tang, C.C., Call, K., Hermanson, G., Evans, G.A., Housman, D. and Ward, D.C. (1990) *Science* **24**, 64-69.

A PstI RFLP of the LACI gene

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Source/Description: A lipoprotein associated coagulation inhibitor gene cDNA fragment containing nucleotides 458 through 1208 (1) cloned in the SmaI site of pUC13.

Polymorphism: PstI digestion reveals a two allele polymorphism with bands at either 6.4 kb or 6.9 kb.

Frequency: Allele frequencies were calculated from 25 unrelated Caucasians.

A1 (6.4 kb) 0.34

A2 (6.9 kb) 0.66

Mendelian Inheritance: Co-dominant inheritance in one family.

Probe Availability: Contact C.P.E.van der Logt.

Reference: 1) Girard *et al.* (1989) *Thromb. Res.* **55**, 37-50.

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