Dinucleotide repeat polymorphism at the D11S436 locus

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Source and Description of Clone: Cosmid cCI11-30, was from a human chromosome 11 specific library (1). A Sau3A subclone of this cosmid (C11G30-S1) was isolated by hybridization to a (CA)₁₀ oligonucleotide and partially sequenced (EMBL accession # X61501). The sequences flanking a (CA)₁₈ repeat were used to design PCR primers.

PCR Primers: C11G30C #1 = 5'-CTCAATCATAGC-AGGGGAC-3'
C11G30C #2 = 5'-CACACCTGGCAATTTGCAA-3'

Polymorphism: Allelic fragments were resolved on DNA sequencing gels.

Allele	Length (nt)	Frequency
C1	188	.024
C2	186	.176
C3	184	.071
C4	182	.023
C5	180	.588
C6	178	.094
C7	176	.024

Alleles in four CEPH parents were as follows: 134001: A2,A2; 134002: A2,A5; 136201: A3,A4; 136202: A5,A5. Allele frequencies were measured in 42 unrelated CEPH parents. Heterozygosity in a sample of 72 unrelated CEPH parents was 47%. When haplotyped with the TaqI RFLP at the D11S436 locus (1), 47 of 69 unrelated CEPH parents (68%) were heterozygous for at least one of the two polymorphisms.

Mendelian Inheritance: Mendelian inheritance was observed in 4 informative CEPH families with a total of 28 children.

Chromosomal Localization: Cosmid cCI11-30 was mapped to 11p12-p11.22 by use of a somatic cell hybrid panel (1).

PCR Conditions: PCR was performed using standard buffer and dNTP concentrations according to (2) except that spermidine was added to 0.25 mM and the products were not radioactively labelled. Primers were present at concentrations of 0.2 μ M. Amplification in a Perkin-Elmer/Cetus System 9600TM thermocycler was for 30 cycles with denaturation at 94°C for 5 sec and annealing/extension at 55°C for 15 sec. PCR products were resolved on DNA sequencing gels, capillary-blotted onto Hybond N+TM nylon membranes (Amersham) and revealed by probing with 5' [³²P] labeled (CA)₁₅ oligomer.

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MspI RFLP of FSHB on chromosome 11p

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Source and Description of Clone: pFSHB-1.1 is a 1.1 kb single-copy EcoRI-SacI human genomic DNA fragment in phage λ .

Polymorphism: MspI identifies two alleles.

Allele B1 - 10.65 kb

Allele B2 - 1.85 kb

Frequencies: In 34 unrelated Caucasian individuals:

Allele B1 (10.65 kb): 0.53

Allele B2 (1.85 kb): 0.47

17/27 (63%) individuals were heterozygous.

Not Polymorphic For: PstI, TaqI, EcoRI, SstI, BamHI, BglII, PvuII, MboI and EcoRV in 9 unrelated Caucasians.

Chromosomal Localisation: FSHB gene has been mapped to human chromosome region 11p11.2-11p13 (2).

Mendelian Inheritance: Mendelian inheritance was demonstrated in six two-generation families (16 meioses scored).

Probe Availability: Contact P.C. Watkins at Integrated Genetics Inc., Framingham.

Other Comments: A HindIII RFLP has previously been detected with this probe (1). In 13 individuals in whom haplotypes could be unequivocally scored, there were 14 HindIII-1/MspI-1, 7 HindIII-2/MspI-2 and 5 HindIII-1/MspI-2 haplotypes. The two RFLPs are thus in strong ($P = 0.0012$) but not perfect disequilibrium.

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