A highly polymorphic VNTR locus on the long arm of chromosome 4

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Source: The probe, EFD 139.1, was isolated as described by Nakamura *et al.* (1). It is cloned into the plasmid vector pUC18. The locus designation is D4S184.

Polymorphism: The probe EFD 139.1 detects a multi-allele polymorphism in HindIII digests of human DNA. At least 11 different alleles have been observed. The allelic fragments vary in size from 3.5 to 8.0 kbp. Three invariant fragments are observed at 0.5, 0.7 and 2.0 kbp.

Chromosomal Localization: The allelic fragments were assigned to the q21-qter region of chromosome 4 using somatic cell hybrids (2). The three constant bands are derived from the acrocentric chromosomes (13, 14, 15, 21 and 22) as determined from a hybrid cell mapping panel.

Mendelian Inheritance: The codominant inheritance of alleles was demonstrated in 30 meioses (see Fig.).

Heterozygosity: In a sample of 20 unrelated Caucasians the heterozygosity was 90% (18/20).

Hybridization Conditions: The entire plasmid was labelled by the random primer method. Repetitive sequences were prevented from hybridizing by preannealing the probe with human DNA. Hybridization was at 65°C for 14 h. Blots were washed three times in 0.5×SSC with 0.1% SDS at 65°C.

Probe Availability: Freely available.

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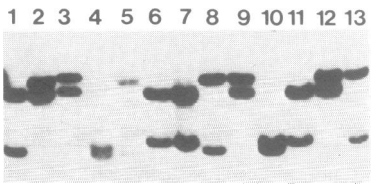
References: 1) Nakamura,Y. *et al.* (1987) *Science* **234**, 1616–1622. 2) Smith,B. *et al.* (1988) *Am. J. Hum. Genet.* **42**, 334–344.

Figure: Segregation of the EFD 139.1 polymorphism. Eleven offspring (lanes 2–12) are flanked by their parents (lanes 1 and 13). The parents are heterozygous for four distinct alleles.



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Dinucleotide repeat polymorphism at the D1S117 locus

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Source and Description: Cosmid ICRFc102H1222 was from a flow sorted chromosome 21 cosmid library. DNA sequences flanking the sequence $(TG)_{21}(TA)_{10}$ within a HaeIII subclone (2H12A) of this cosmid (EMBL accession # X54558) were used to design PCR primers.

PCR Primers:
2H12A # 1: 5′-CCTTTTGCCTCCTTCGT-3′
2H12A # 2: 5′-CTCATTTACAATAGCTACC-3′.

Polymorphism: Eleven allelic fragments were resolved on DNA sequencing gels. Lengths of allelic fragments (nt) were: A1 = 132, A2 = 126, A3 = 124, A4 = 122, A5 = 120, A6 = 118, A7 = 116, A8 = 114, A9 = 112, A10 = 110, A11 = 100.

Frequencies: Allele frequencies in 28 unrelated European Caucasians were:

A1	= .018	A7	= .018
A2	= .036	A8	= .036
A3	= .054	A9	= .071
A4	= .125	A10	= .036
A5	= .357	A11	= .036
A6	= .214	PIC	= 0.77

Chromosomal Localization and Mendelian Inheritance: PCR of genomic DNAs from a somatic cell hybrid panel indicated localization to chromosome 1. Linkage analysis in 4 CEPH families with 28 informative meioses against locus AT3 (localized to q23–q25.1 (1)), gave a maximum LOD score of 5.06 at theta = 0.065. Mendelian inheritance was observed in all cases.

PCR Conditions: PCR reactions are carried out in total volume of 25 μl containing: 25 ng genomic DNA, 10 pmole of each primer, 1.5 mM $MgCl_2$, 200 μM dNTPs, 50 mM KCl, 10 mM tris-Cl, pH 8.3, 0.6 units Taq polymerase (BRL) and 0.01% gelatin. Amplification is for 35 cycles with denaturation at 94°C for 60 seconds, annealing at 46°C for 60 seconds and extension at 72°C for 30 seconds.

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Reference: 1) Bruns,G.A.P. and Sherman,S.L. (1989) *Cytogenet. Cell Genet.* **51**, 67–90.

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