Highly polymorphic RFLP marker D4S130 (CRI-L518) maps to distal 4q

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Source/Description: D4S130 was isolated from a genomic lambda charon 4A library (1). Insert size is 10–15 kb.

Polymorphism: D4S130 detects a multi-allele polymorphism with HaeIII digests of human DNA; over 14 alleles have been seen. Variable fragments range in size from 1.0 to 3.5 kb. Constant bands are observed at 2.6, 2.0, 1.3, 0.98, 0.82, and 0.74 kb. The probe detects two systems with TaqI digested human DNA. Polymorphisms are also detected with BglII, EcoRI, MspI, PstI, and RsaI. The Taq and HaeIII polymorphisms are in linkage disequilibrium.

Heterozygosity: Using the parents of 21 CEPH families, the observed heterozygosity for the HaeIII polymorphism was 0.71 and the PIC was 0.69.

Frequency: The following frequencies of HaeIII alleles were seen in 18 CEPH parents

Allele	Size (kb)	Frequency	Allele	Size (kb)	Frequency
C1	3.1	2.8% (1/36)	C8	1.85	2.8% (1/36)
C2	2.95	5.6% (2/36)	C9	1.6	11.1% (4/36)
C3	2.9	2.8% (1/36)	C10	1.55	5.6% (2/36)
C4	2.8	11.1% (4/36)	C11	1.38	8.3% (3/36)
C5	2.2	2.8% (1/36)	C12	1.35	11.1% (4/36)
C6	2.1	22% (8/36)	C13	1.26	5.6% (2/36)
C7	2.05	2.8% (1/36)	C14	1.2	5.6% (2/36)

Not Polymorphic For: BamHI and HindIII.

Chromosomal Localization: D4S130 was provisionally assigned to chromosome 22 based on a two-point lod score greater than 3 but less than 4 (2). It was later reassigned to chromosome 4 by a somatic cell hybrid panel (3) but was unlinked to most chromosome 4 markers. We now find that HaeIII RFLP detected by D4S130 is tightly linked to distal chromosome 4q markers D4S139 (4q35), D4S184, and D4S171 using 40 CEPH reference families.

Interval	Sex-equal Recombination Fraction	Maximum Lod
D4S130-D4S139	0.02	141.79
D4S130-D4S184	0.01	134.35
D4S130-D4S171	0.11	64.17

Mendelian Inheritance: Co-dominant inheritance of the HaeIII RFLP was seen in the 36 multigenerational CEPH families.

Probe Availability: D4S130 is available for research use. Contact G.Vovis, Collaborative Research, Inc.

References: 1) Lawn *et al.* (1978) *Cell* **15**, 1151–1174. 2) Donis-Keller *et al.* (1987) *Cell* **51**, 319–337. 3) Donis-Keller *et al.* (1989) *Cytogenet. Cell Genet.* **51**, 991–992.

A PvuII polymorphism detected by pEW404 (D17S64) on chromosome 17

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Source and Description: pEW404 (D17S64) is a 1.4 kb genomic fragment subcloned into plasmid pUC8.

Polymorphism: PvuII detects a two allele polymorphism with fragments of 13.0 B1 and 7.8 B2 kb.

Frequency: Estimated from 42 unrelated Caucasians

13.0 kb (B1) = .4

7.8 kb (B2) = .6

Observed heterozygosity is 0.43

Not Polymorphic For: BamHI, BglI, BglII, EcoRI, EcoRV, HaeIII, HincII, HindIII, HinfI, KpnI, NciI, RsaI, SacI, SstI, StuI, TaqI, XbaI (studied in 6 unrelated individuals).

Chromosomal Location: 17p11.2–17p12.0 (1).

Mendelian Inheritance: Co-dominant segregation was observed in 6 families.

Probe Availability: Available from ATCC.

Other Comments: RFLP was observed under hybridization conditions using preannealing with Human Placental DNA and very stringent washes. Linkage disequilibrium was not observed between the MspI (1) and PVUII polymorphisms.

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References: 1) Solomon, E. and Barker, D.F. (1989) *Cytogenetics and Cell Genetics* **51**, 319–337.

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