A new *Sty*I RFLP and haplotypes with the *Hin*dIII RFLP at the D8S5 (TL11) locus

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Description, Source and Method: Probe TL11 (locus D8S5) is an anonymous 9.3 kb insert in lambda phage isolated from a human genomic library (1). Standard Southern gel blotting in 0.8% agarose gels detects from 4 to 6 discrete restriction fragments if the probe is preassociated with sheared, boiled human placental DNA.

Variation and Other Enzymes: *Hin*dIII detects a previously reported RFLP (1); we report a new RFLP detected by *Sty*I (Figure). Testing a minimum of 9 unrelated Caucasians, TL11 is not polymorphic for *Eco*RI, *Bam*HI, *Msp*I, *Taq*I, *Pst*I (1); and *Apa*I, *Bcl*I, *Bgl*I, BglII, *Dra*I, *Hgi*AI, *Mbo*I, *Nsi*I, *Rsa*I, *Sac*I and *Xho*I.

Inheritance and Frequency: Mendelian inheritance was observed in 40 CEPH (Paris) reference families. Allele and haplotype frequencies in 74 unrelated CEPH individuals are:

enzyme	allele (kb)	freq.	haplotypes	obs. freq.	exp. freq.	heterozygosity	
*Sty*I	8.1	.18	8.1−11.8	.18	.13	*Sty*I	.30
	4.5	.82	8.1−10.6	.00	.05	*Hin*dIII	.39
*Hin*dIII	11.8	.73	4.5−11.8	.55	.60	*Sty*I−*Hin*dIII (obs.)	.55
	10.6	.27	4.5−10.6	.27	.22	*Sty*I−*Hin*dIII (exp.)	.58

The standardized disequilibrium for these haplotypes, $\Delta = -.287$, is significantly different from 0 ($p < .01$) but the observed and expected heterozygosities are very similar. The 8.1−10.6 haplotype has been observed in other Caucasian families (unpublished).

Chromosomal Location: Earlier localized to proximal 8q but more recently localized to 8p23-q11 (2). Linkage mapping is consistent with localization to 8p (3 and unpublished).

Availability: Available from Dr. Warnich or from the American Type Culture Collection (ATCC).

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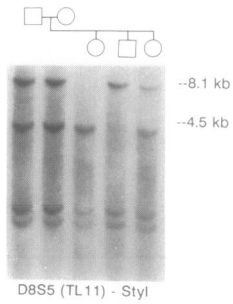

Dinucleotide repeat polymorphism at the D11S527 locus

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Source and Description of Clone: Cosmid 8G11, also known as c23,23 or cosmid ZD5 was from a human chromosome 11q specific library (1). A subclone of this cosmid (pJG4) was isolated by hybridization to a $(CA)_{15}$ oligonucleotide and partially sequenced. The sequences (EMBL accession number: X60166) flanking the repeat $(GT)_{17}(CTGT)_6$ were used to design PCR primers.

PCR Primers:
JG4-A = 5′-GCCCCTCTACTTGTCTGGAG-3′
JG4-C = 5′-ATGCGGCTCCAAGACAAGTTC-3′

Polymorphism: Allelic fragments were detected on DNA sequencing gels. Lengths (nt) are: A1 = 166, A2 = 164, A3 = 162, A4 = 160, A5 = 158, A6 = 156, A7 = 154, A8 = 152, A9 = 150, A10 = 148, A11 = 146, A12 = 142. Alleles in four CEPH parents were as follows: 134101: A4,A9; 134102: A10,A10; 141801: A4,A9; 141802: A3,A11.

Frequencies: from 77 unrelated CEPH parents: A1 = 0.013, A2 = 0.11, A3 = 0.097, A4 = 0.156, A5 = 0.162, A6 = 0.058, A7 = 0.136, A8 = 0.019, A9 = 0.091, A10 = 0.078, A11 = 0.071, A12 = 0.006. The PIC calculated from these frequencies is 0.88.

Mendelian Inheritance: Mendelian inheritance was observed in 10 informative CEPH families with a total of 79 children.

Chromosomal Localization: Cosmid ZD5 was mapped to 11q13.5 by fluorescent in situ hybridization (1). Three-point linkage analysis of 10 informative CEPH families suggests the order cen-D11S97-D11S527-D11S388-qter, which is consistent with the in situ hybridization data. Odds against inversion of D11S527 and D11S97 are 100:1; odds against inversion of D11S527 andD11S388 are 3800:1.

PCR Conditions: PCR was performed according to (2) except that the products were not radioactively labelled. Annealing temperature was 60°C. PCR products were resolved on DNA sequencing gels, capillary-blotted onto Hybond N+™ nylon membranes (Amersham) and revealed by probing with 5′[^{32}P] labeled $(CA)_{15}$ oligomer.

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