A new RFLP marker D5S348 maps to 5p14.3 – 15.2, between D5S60 (CRI-R535) and HPRTP2

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Source/Description: JH-6 (D5S348) a 14.7 kb insert cloned into the *Bam*HI site of λ DASH II was derived from a human YAC clone B24C12 (Washington University).

Polymorphism:

Enzyme	Allele	Size	Constant Bands
<i>Bgl</i> III	A1	13.0 kb	5.2 kb
	A2	12.0 kb	1.4 kb

Frequencies: Determined from parents in 40 CEPH families.

Allele	Frequency	PIC	Heterozygosity(%)	Informative Meioses
A1	0.65	0.35	45	204
A2	0.35			

Not Polymorphic For: *Bam*HI, *Eco*RI, *Hind*III, *Hinc*II, *Msp*I, *Pst*I, or *Taq*I (6 unrelated individuals tested).

Mendelian Inheritance: Co-dominant inheritance observed in CEPH reference pedigrees.

Chromosomal Localization: FISH localised JH-6 to 5p14.3–15.2. Genetic linkage analysis using genotype data collected from CEPH pedigrees, the CEPH V5 database, and the program package CRI-MAP (1) confirms this localization. Figure 1 shows a sex-averaged map (odds for orders $\geq 1000:1$).

Probe Availability: From the ATCC (cat. nos. 65832, 65833).

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References: 1) Donis-Keller,H. *et al* (1987) *Cell* **51**, 319–337.

Table 1. Two-point LOD table of D5S348 (JH-6) versus chromosome 5p markers

Probe	Locus	Zmax	θ
CRI-C44	D5S56	11.11	0.14
CRI-L433	D5S59	12.03	0.08
CRI-R535	D5S60	13.38	0.01
plambda500	HPRTP2	5.71	0.15
CRI-1072	D5S69	6.06	0.15

Figure 1. Regional map of chromosome 5p.

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A *Hinf* I polymorphism in the human elastin gene (ELN)

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Source/Description: Human elastin (ELN) genomic *Eco*R I restriction fragments were isolated from the elastin clone HEL-6 (1), for use as a probe.

Polymorphism: This probe detects a two allele system in human genomic DNA restricted with *Hinf* I and subjected to Southern analysis (Fig.1). Allele one (D1) two 750bp fragments, allele two (D2) a 1.5kb fragment.

Frequency: Estimated in 31 unrelated healthy British Caucasians. D1 = 0.61; D2 = 0.39. Of 31 individuals there were 14 heterozygotes, 12 homozygotes (D1) and 5 homozygotes (D2). PIC = 0.363.

Major constant bands are detected at 2.9, 2.0 and 1.7 kb.

Chromosomal Location: The elastin gene has been localised to 7q11.2 (2).

Mendelian Inheritance: Mendelian inheritance was observed in three families (nine individuals).

Other Comments: High background signal in Southern analyses, caused by repetitive elements in the genomic probe, was suppressed by the use of human genomic DNA sonicated to 500 bp.

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Figure 1. Autoradiograph of a *Hinf* I polymorphism in normal human DNA samples probed with the genomic clone HEL-6 : (a) heterozygote, (b and c) homozygotes A1, (d) homozygote A2.