Isolation of a polymorphic DNA sequence (LL101) from the short arm of chromosome 17 [D17S251]

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Source/Description: LL101 is a recombinant bacteriophage (vector: λ Ch3A Δ lacXN) containing a 9.0kb *NotI* fragment. This clone was isolated from a *NotI* linking library constructed from the chromosome 17 fraction of a flow-sort of human chromosomes (1).

Polymorphism: *PvuII* identifies a two-allele polymorphism with fragments of 13kb (P1) and 8.6kb (P2) in a Caucasian population. Three additional alleles were observed in U.S. blacks (P3 = 15kb; P4 = 8kb; P5 = 6kb). Invariant fragments of 2.0kb, 1.6kb, 1.1kb, 1.0kb, 0.64kb and 0.5kb are also observed.

Allele Frequency:	P1	P2	P3	P4	P5
U.S. Blacks (N = 6)	0.49	0.16	0.08	0.08	0.16

Not Polymorphic For: *MspI*, *PstI*, *RsaI*.

Chromosomal Localization: LL101 is localized to 17p13.100–17p13.105 using a previously published chromosome 17 somatic cell hybrid panel (2, 3). This localization is based on the presence of LL101 in hybrid JW-4 retaining a del(17)(p13.105) (2) and its absence in hybrid HO-11 retaining a del(17)(p13.100) (3).

Mendelian Inheritance: Co-dominant segregation of the two-allele polymorphism (P1, P2) has been observed in one three generation family where 34 individuals were typed (4).

Probe Availability: Contact P.I.Patel.

Other Comments: Preassociation with human placental DNA is required prior to hybridization at normal stringency (65°C, 1MNa⁺).

References: 1. Wallace, M.R. *et al.* (1989) *Nucl. Acids Res.* **17**, 1665–1677. 2. van Tuinen, P. *et al.* (1987) *Genomics* **1**, 374–381. 3. van Tuinen, P. *et al.* (1988) *Am. J. Hum. Genet.* **43**, 587–596. 4. Patel, P.I. *et al.* (1990) *Am. J. Hum. Genet.* **46**.

MspI and *PvuII* polymorphisms in the Na,K-ATPase β subunit gene ATP1B1

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Source/Description: ATP1B HH1.2 is a 1.2 kb *HindIII* fragment from the 3' portion of the human Na,K-ATPase β subunit gene, ATP1B1 (1).

Polymorphism: *MspI* identifies a two allele polymorphism (M1: 6.7 kb, M2: 5.3 kb). *PvuII* also detects a two-allele polymorphism (P1: 5.1 kb, P2: 4.7 kb).

Frequency: Estimated from 15 unrelated Caucasians.

<i>MspI</i> M1: 0.5	<i>PvuII</i> P1: 0.63
M2: 0.5	P2: 0.37

Not Polymorphic For: *BamHI*, *BglII*, *HindIII*, *KpnI*, *PstI*, *SstI*, *XbaI*.

Chromosome Location: ATP1B1 has been assigned to chromosome 1q by somatic cell hybrid analysis (2).

Mendelian Inheritance: Codominant segregation of the *MspI* RFLP was observed in one two-generation family (5 individuals). Codominant segregation of the *PvuII* RFLP was observed in a two-generation (8 individuals) and a three-generation (12 individuals) family. Linkage disequilibrium exists between the two polymorphisms. Although insufficient chromosomes were investigated to compute the level of disequilibrium accurately, it is evident that added information will be gained by using both polymorphisms.

Probe Availability: Contact Dr J.B.Lingrel.

References: 1. Lane, L.K. *et al.* (1989) *Genomics* **5**, 445–453. 2. Yang-Feng, T.L. *et al.* (1988) *Genomics* **2**, 128–138.

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