MspI and PvuII polymorphisms in the Na,K-ATPase α subunit related gene ATP1A1

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Source/Description: ATP1A1 78-1-3 is a 0.56 kb genomic *EcoRI-XbaI* fragment from within the Na,K-ATPase α subunit related gene, previously referred to as α D (1), on chromosome 13 (2). The fragment was subcloned into pIBI31.

Polymorphism: *MspI* identifies a two-allele polymorphism (M1: 2.8 kb, M2: 2.5 kb). *PvuII*, which cuts within the probe sequence, detects two two-allele polymorphisms (A1: 6.0 kb, A2: 5.7 kb, B1: 1.3 kb, B2: 1.1 kb). A1 and A2 appear to result from an insertion/deletion polymorphism that is also identified by *MspI*.

Frequency: Estimated from 15 unrelated Caucasians.

<i>MspI</i>	M1: 0.7	<i>PvuII</i>	A1: 0.7
	M2: 0.3		A2: 0.3
			B1: 0.3
			B2: 0.7

Not Polymorphic For: *BamHI*, *BglII*, *EcoRI*, *HindIII*, *KpnI*, *PstI*, *SacI*, and *XbaI*.

Chromosomal Location: ATP1A1 78-1-3 has been assigned to chromosome 13q by somatic cell hybrid analysis (2).

Mendelian Inheritance: Codominant segregation of the RFLPs was observed in 2 two-generation families.

Probe Availability: Contact Dr J.B.Lingrel.

References

1. Shull, M.M. and Lingrel, J.B. (1987) *Proc. Natl. Acad. Sci. USA* **84**, 4039-4043.
2. Yang-Feng, T.L. et al. (1988) *Genomics* **2**, 128-138.

A new polymorphic probe on chromosome 3p: λ LIB28-77 (D3S169E)

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Source/Description: λ LIB28-77 is a 4.0 kb *EcoRI* fragment isolated from a Charon 21A human chromosome 3 library (1).

Polymorphism: *Taq I* digestion of genomic DNA and hybridization with this probe detected a two allele polymorphism: 19.0 kb (T1) and 12.5 kb (T). An 0.6 kb constant band is present.

Frequency: Estimated from 40 unrelated Caucasians.

T1	: 0.15
T2	: 0.85

Not Polymorphic For: *Hind III*, *Msp I*, *Pst I*, *Rsa I*.

Chromosomal Localization: Using a somatic cell hybrid panel (2) which is based on linkage groups anchored to physically localized markers, the probe was assigned to the short arm of chromosome 3 (3p).

Mendelian Inheritance: Mendelian inheritance has been demonstrated in a two-generation von Hippel-Lindau disease (VHL) family.

Probe Availability: Available for collaboration.

Other Comments: The probe consists of unique sequences which were cDNA selected and are highly conserved between human, rodent and murine species.

- References:** 1. American Type Culture Collection No. 57748, 2. Brauch, H. et al: Human Gene Mapping 10, Abstract No. A2212, 1989 (in press).

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