TaqI polymorphism in the 3' flanking region of the human aortic smooth muscle actin gene

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Source and Description of Clone: A 2 kb *EcoRI* fragment from the 3' flanking region of the human aortic smooth muscle actin gene that was subcloned into pUC18¹ (pASMD_e).

Polymorphism: *TaqI* identifies two alleles with DNA fragments at 8.5 kb or at 5 kb and 3.5 kb.

Frequency: Unrelated individuals (26 Japanese and 5 Caucasians) studied: 24 males, 7 females (62 chromosomes).

8.5 kb allele 0.81
5 kb and 3.5 kb allele 0.19

Not Polymorphic For: *BanI*, *BanII*, *BglI*, *DraI*, *EcoRI*, *HindIII*, *MspI*, *NcoI*, *PstI*, *PvuII*, *ScaI*, *XbaI*.

Chromosome Localization: 10q22 to 10qter².

Mendelian Inheritance: Co-dominant segregation in five families.

Probe Availability: Available for collaboration.

Other Comments: Low background under usual stringency conditions. Final wash at 0.1×SSC at 65°C.

References: 1. Kamada, S. *et al.* (1989) *Gene*, in press. 2. Ueyama, H. *et al.* in preparation.

Scrfl restriction fragment length polymorphism at the D7S23 locus (probe pKM.19), closely linked to cystic fibrosis

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Source/Description: pKM.19 is a 1.0 kb *EcoRI* genomic fragment in pUC13 (ref. 1, 2). pP1 was isolated independently but contains the same fragment as pKM.19 (ref. 3).

Polymorphism: *Scrfl* (CC/NGG) identifies a two allele polymorphism with bands at 0.86 kb (A1) and 0.61 + 0.27 kb (A2), and a constant band of 0.52 kb.

Frequency:		CF		non-CF	
Population	n	A1	A2	n	A1 A2
Spanish	80	0.63	0.36	80	0.04 0.96
British	32	0.78	0.22	31	0.06 0.94

Not Polymorphic For: *BamHI*, *HindIII*, *PvuII*, *XbaI*, *EcoRI*, *TaqI*, *HincII*.

Chromosomal Localisation: To chromosome 7q31 by linkage analysis and using a panel of somatic human/rodent cell hybrids (1, 2).

Mendelian Inheritance: Co-dominant segregation was demonstrated in 100 families.

Probe Availability: Freely available from X. Estivill.

Other Comments: The same probe detects a *PstI* polymorphism which was previously described (2).

References: 1. Estivill *et al.* (1987) *Nature* **326**, 840-845. 2. Estivill *et al.* (1987) *Genomics* **1**, 257-263. 3. Ramsay *et al.* (1990) *Genomics* in press.

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