

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

Hisao Ueyama and Rie Ohsugi
Department of Medical Biochemistry, Shiga University of Medical Science, Seta, Otsu 520-21, Japan

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 (pASMDc).

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: BamI, BamII, 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.

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

V.Nunes, M.Ramsay¹, T.Casals, M.Chillón, N.Lench¹, M.Schwartz² and X.Estivill
Molecular Genetics Department, Fundació d'Investigació Sant Pau, Barcelona, Spain, ¹Molecular Genetics Department, St Mary's Hospital Medical School, London and ²Clinical Genetics Unit, Royal Manchester Children's Hospital, Manchester, UK

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: ScrfI (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		
	n	A1	A2	n	A1	A2
Population	80	0.63	0.36	80	0.04	0.96
Spanish	32	0.78	0.22	31	0.06	0.94
British						

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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