

**RFLPs at the D21S19 locus of human chromosome 21**

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**SOURCE/DESCRIPTION:** Probe GMG21S5:- a 6.4kb single-copy Eco RI fragment isolated from a flow sorted, chromosome 21-specific DNA library and sub-cloned into PUC9 to form plasmid pGSB3. (see ref. 1)

**CHROMOSOMAL LOCATION:** 21q21.2 - 21qter by somatic cell hybrid and by flow sorting. (1)

**POLYMORPHISMS:** Recognises two, two allele polymorphisms when used to probe human genomic DNA digested with Pst I (shown in table below). There is one constant band at 1.2kb. Additional polymorphisms recognised by Apa I, Bam HI, Msp I and Nci I are in disequilibrium with the upper Pst I polymorphism and a Ban II polymorphism is in disequilibrium with the lower one. The two Pst I polymorphisms are not in complete disequilibrium with each other in the Venezuelan population examined.

Pst I Polymorphisms		Size	Frequency (29 Individuals)
Upper	A1	3.6kb	0.04
	A2	2.2kb	0.96
Lower	A1	1.65kb	0.8
	A2	1.5kb	0.2

**NOT POLYMORPHIC FOR:** Ban I, Bcl I, Bgl I, Bgl II, Bsp 1286, Bst E2, Bst NI, Bst XI, Eco RI, Eco RV, Hae II, Hinc II, Hind III, Hinf I, Hph I, Kpn I, Mbo I, Nde I, Pvu II, Rsa I, Sac I, Sau 96-I, Sca I, Scr FI, Sst I, Stu I, Taq I, Tth I, Xba I and Xmn I when tested against 5 unrelated individuals.

**MENDELIAN INHERITANCE:** Co-dominant segregation in 12 informative families.

**COMMENTS:** Due to the disequilibrium all informative matings can be assayed by using Pst I. A 60V, 1% agarose gel run overnight in TBE is sufficient to resolve the alleles. The gel can be cut 3cm below the wells. Additional bands may be observed at low stringencies.

**PROBE AVAILABILITY:** Available on request.

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**REFERENCE:** (1) Stewart, G.D., Harris, P., Galt, J. and Ferguson-Smith, M.A. (1985) Nucl. Acids Res. 13:4125-4132.