Two polymorphisms at the DXS539 locus

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Source/Description: pJH89 is a 2.3 kbp *SalI/PstI* fragment isolated from a phage library derived from a reduced human-hamster somatic cell hybrid (908K1B17) and subcloned in pGEM3 (1).

Polymorphism: pJH89 detects two two-allele polymorphisms:

<i>TaqI</i>	2.6 kbp	A1
	2.2 kbp	A2
<i>NcoI</i>	5.5 kbp	B1
	4.6 kbp	B2

With *TaqI* constant bands of 0.9 kbp, 0.4 kbp, and 0.2 kbp are seen and with *NcoI* constant bands of 2.4 kbp and 2.7 kbp are seen.

Frequency:	Allele	Frequency	Number of Chromosomes
	A1	0.16	160
	A2	0.84	
	B1	0.83	88
	B2	0.17	

Not Polymorphic For: *BamHI*, *BclI*, *BglI*, *BglII*, *EcoRI*, *EcoRV*, *HindIII*, *KpnI*, *MspI*, *PstI*, *PvuII*, *AvaII*, *SstI*, and *XbaI*.

Chromosomal Localisation: Proximal but closely linked to FRAXA.

Mendelian Inheritance: X-linked codominant segregation was observed in more than 20 Fragile-X families.

Probe Availability: Write to B.A.van Oost.

Other Comments: The two polymorphisms are almost in equilibrium as the observed combined heterozygosity is 42%. Genetic and physical mapping have demonstrated that DXS539 is located in between DXS369 and FRAXA (Dreesen *et al.*, manuscript in preparation).

Reference: 1) Schonk, D. *et al.* (1989) *Genomics* 4, 383–396.

EcoRI and PvuII RFLPs in the endonexin II/annexin V (ANX5) gene on chromosome four

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Source/Description: pRK6 contains a 1.3 kb *NcoI/HindIII* cDNA insert cloned from human placenta. Sequence of the insert extends from the *NcoI* site at the ATG to the *HindIII* site 323 bp downstream of the translational stop codon. The vector is pKK233–2 (1).

Frequency:

	Allele	band size (kb)	frequency
		18 13 12 7	
		18 13 12 7	
EcoRI	A1	+ + - +	.66 (56 CEPH chromosomes)
	A2	+ - + +	.34 (heterozygosity = .61)
		12 7 6.2 4.8 3.5	
PvuII	B1	+ + - - +	.44 (134 CEPH chromosomes)
	B2	- + + + +	.56 (heterozygosity = .49)

Not Polymorphic For: *AccI*, *AvaI*, *BamHI*, *BanI*, *BanII*, *Eco109*, *EcoRV*, *HincII*, *HindIII*, *KpnI*, *MspI*, *RsaI*, *SspI*, *StyI*, *XbaI*, *XhoI*.

Chromosomal Localisation: 4q28–q32 (2).

Mendelian Inheritance: Co-dominant segregation was seen in 35 CEPH families.

Probe Availability: Contact M.Jaye.

Other Comments: Strong linkage disequilibrium was observed between the two RFLPs.

References: 1) *J. Biol. Chem.* 263, 8037–8042 (1988). 2) *Cytogenet. Cell Genet.* 52, 167 (1989).

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