An anonymous DNA probe E20 [D4S143] on chromosome 4 detects a *TaqI* polymorphism

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Source and Description: A 13kb fragment cloned into the *EcoRI* site of phage lambda L47.1, isolated from a human genomic library.

Polymorphism: *TaqI* identifies a two allele polymorphism (A1: 4.7kb; A2: 7.2 kb) with constant fragments at 1.75 and 2.15kb.

Frequency: Studied in 79 parents of CEPH families.

A1: 0.5570

A2: 0.4430

Not Polymorphic for: *EcoRI*, *HindIII*, *BamHI*, and *PstI* with DNA of 5 unrelated individuals.

Chromosomal Localization: The probe was localised to 4q21–24 by means of *in situ* hybridization.

Mendelian Inheritance: Co-dominant segregation has been observed in 21 informative CEPH families.

Probe Availability: Available for collaboration.

Other Comments: The probe was pre-associated with an excess of sonicated total human DNA prior to hybridization.

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A *BclI* RFLP for *DXS296* (VK21) near the fragile X

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Source/Description: Four single copy plasmid sub-clones of VK21:VK21A (1.3 kb), VK21B (2.2 kb), VK21C (2.5 kb), and VK21D (3.2 kb) were used as probes.

Polymorphism: *BclI* (New England Biolabs) detected a polymorphism with VK21A, in addition to previously reported polymorphisms identified by VK21A with *TaqI* and VK21C with *MspI* (Suthers, *et al.*, 1989).

<i>TaqI</i>	10.9 kb A1	<i>BclI</i>	10.7 kb C1
	9.9 kb A2		5.0 kb C2
<i>MspI</i>	12.7 kb B1	(present study)	
	9.9 kb B2		

(Suthers *et al.*, 1989)

Frequency: From 27 unrelated individuals (total 45X chromosomes): C1=0.87, C2=0.13. All haplotypes were either A1B1C1 or A2B2C2.

Not Polymorphic for: *BanI*, *BglI*, *BstNI*, *BstXI*, *EcoRV*, *XbaI*, *XmnI* in addition to previously reported restriction enzymes (Suthers *et al.*, 1989).

Chromosomal Localization: Distal but closely linked to FRAXA (Suthers *et al.*, 1989).

Mendelian Inheritance: Codominant inheritance was demonstrated in Utah Families 981 and 983. In both families the male parent was hemizygous for the C1 allele and the female parent heterozygous C1C2. From a total of 15 offspring there were 7 males hemizygous for the C2 allele, 4 males hemizygous for the C1 allele, 2 females heterozygous C1C2 and 2 females homozygous C1C1.

Probe Availability: Available on a collaborative basis from Dr.G.R.Sutherland, Cytogenetics Unit, Department of Histopathology, Adelaide Children's Hospital, North Adelaide, S.A. 5006, Australia.

Other Comments: The VK21 probes are useful for diagnosis in families with the fragile X. This extended search for RFLPs was undertaken in an attempt to increase their informativeness. Although the new *BclI* marker appears to be in linkage disequilibrium with the existing *TaqI* and *MspI* markers, the availability of a *BclI* RFLP adds flexibility to diagnostic strategies based on reprobings of filters since *BclI* also defines other markers near FRAXA: *DXS52* (F814 and St-14), *DXS105* (55.E), and *DXS304* (U6.2).

Reference: G.K.Suthers, D.F.Callen, V.J.Hyland, H.M.Kozman, E.Baker, H.Eyre, P.S.Harper, S.H.Roberts, M.C.Hors-Cayla, K.E.Davies, M.V.Bell and G.R.Sutherland. A new DNA marker tightly linked to the fragile X locus (*FRAXA*). (1989) *Science* **246**, 1298–1300.