Three RFLPs for pZ11 (DXS540) in the choroideremia gene at Xq21.2

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Source and Description: pZ11a/b (DXS540) consists of 1.2 and 4.2 kb *EcoRI* fragments which are located next to each other in the middle of the choroideremia gene at Xq21.2. pZ11a (1.2 kb *EcoRI*) is a single-copy fragment. pZ11b (4.2 kb *EcoRI*) contains a repetitive sequence. pZ11c is a 1.6 kb *EcoRI/Sau3AI* fragment of pZ11b subcloned in pUC18 (*EcoRI/BamHI*). pZ11 was isolated from a lambdaZAP library which was enriched for sequences from a 625 kb *SfiI* fragment known to span the choroideremia gene (1).

Polymorphisms: pZ11a identifies two-allele polymorphisms with *EcoRV* and *MspI*, pZ11c identifies a two-allele polymorphism with *EcoRI*.

Probe	REN	Size(kb)	Allele	Frequency	X-chromosomes
pZ11a	<i>EcoRV</i>	7.3	B1	0.53	62
		5.3	B2	0.47	
pZ11a	<i>MspI</i>	7.5	C1	0.69	45
		16.5	C2	0.31	
pZ11c	<i>EcoRI</i>	4.5	A1	0.64	62
		4.2	A2		0.36

Not Polymorphic For: *BamHI*, *BglII*, *TaqI*, *HindIII* and *PstI* with pooled DNA of 5 unrelated females.

Chromosomal Localization: The probe was localized to Xq21.2 by means of deletion analysis (1,2).

Mendelian Inheritance: X-linked segregation of the polymorphic alleles was observed in 3 Caucasian families.

Probe Availability: Available on a collaborative basis from F.P.M.Cremers.

References: 1) van de Pol, T.J.R. *et al.* (1990) *Nucleic. Acids. Res.* **18**, 725–731. 2) Cremers, F.P.M. *et al.* (1990) *Am. J. Hum. Genet.* **47**, 622–628.

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