An MspI polymorphism for the HOX2F gene

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Source/Description: pH2-1-3Bg2 is a 1.2 kb BglII fragment subcloned into the BamHI site of Bluescript ks + vector (Stratagene). It was derived from pH2-1-3, a 5.6 kb BamHI fragment from the cosmid clone cH2-1 isolated from a human genomic library (T. Ogura) using the HOX2A clone pBS1 (T. Miki). The clone pH2-1-3 contains the HOX2F homeobox domain by homology of the restriction maps of cosmid cH2-1 with those of Acampora *et al.* (1). This was confirmed by sequencing this homeobox.

Polymorphism: With pH2-1-3Bg2 MspI detects a two allele polymorphism with allelic bands at either 2.0 kb or 1.8 kb. There is a faint constant band at 1.0 kb.

Frequency: Allele frequencies are based on 49 unrelated individuals in four large families of mixed European ancestry and 63 of the unrelated European CEPH parents (i.e. Venezuelans and related parents excluded).

Allele	Allele Specific Band	Kidd Lab 2N = 98	Caucasian CEPH Parents 2N = 126	Combined (2N = 224)
A1	2.0 kb	.70 ± .05	.59 ± .04	.64 ± .03
A2	1.8 kb	.30 ± .05	.41 ± .04	.36 ± .03

Not Polymorphic For: BamHI, BanI, BanII, BclI, BglI, BglII, BstEII, BstNI, DraI, EcoRI, EcoRV, HaeIII, HincII, HinfI, KpnI, PstI, PvuII, RsaI, SacI, XbaI, XmnI using the larger 5.6 kb clone pH2-1-3 on a panel of 8 unrelated Caucasians of mixed European ancestry.

Chromosomal Location: 17q21-q22 based on location of HOX2A (2). Linkage data show no crossovers with the SacI RFLP identified by clone BS3 (3).

Probe Availability: The pH2-1-3Bg2 clone is being sent to ATCC.

Other Comments: A low frequency TaqI variant was noted in three separate families using the 5.6 kb pH2-1-3 clone and is detectable with pH2-1-3Bg2 as a band at 3.4 (freq. 97%) or a band at 2.2 kb (freq. 3%).

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References: 1) Acampora, D. *et al.* (1989) *Nucl. Acids Res.* **17**, 10385-10402. 2) Solomon, E. and Barker, D. (1989) *HGM10: Cytogenet. Cell Gen.* **51**, 319-337. 3) Murphy, P.D. *et al.* (1987) *Nucl. Acids Res.* **15**, 6311.

Two RFLPs at the HOX2G locus

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Source/Description: pH2-1-9 is a 1.8 kb BamHI fragment subcloned into the BamHI site of the bluescript ks + vector (Stratagene) from a cosmid clone, cH2-1, isolated from a human genomic library (T. Ogura) using the HOX2A clone pBS1 (T. Miki). pH2-1-9 maps to the first intron of HOX2G close to HOX2F by homology of the restriction maps of cosmid cH2-1 with those of Acampora *et al.* (1).

Polymorphism:

A TaqI Detects a two allele polymorphism with allelic bands at either 3.8 kb or 3.2 kb. There is a constant band at 1.8 kb.

B MspI Detects a two allele polymorphism with allelic bands at 3.9 kb or at 2.0 kb. There is no constant band.

Frequency: Allele frequencies are based on unrelated individuals in four large families of mixed European ancestry in addition to the unrelated European CEPH parents (i.e. Venezuelans and related parents excluded). Strong, but not complete, linkage disequilibrium exists.

Enzyme	Allele	Allele Specific Bands	Kidd Lab Families	Caucasian CEPH 2N Parents	Combined 2N	2N
TaqI	A1	3.8 kb	.13 ± .04	64 .15 ± .03	136 .14 ± .03	200
	A2	3.2 kb	.87 ± .04	.85 ± .03	.86 ± .03	
MspI	B1	3.9 kb	.97 ± .02	74 .78 ± .04	142 .84 ± .03	216
	B2	2.0 kb	.03 ± .02	.22 ± .04	.16 ± .03	
Haplotypes	A1 B1		.00	60 .05 ± .02	136 .03 ± .01	196
	A1 B2		.07 ± .03	.10 ± .03	.09 ± .02	
	A2 B1		.91 ± .04	.72 ± .04	.78 ± .03	
	A2 B2		.02 ± .02	.13 ± .03	.10 ± .02	

Not Polymorphic For: BamHI, BanI, BanII, BclI, BglI, BglII, BstEII, BstNI, DraI, EcoRI, EcoRV, HaeIII, HincII, HinfI, KpnI, PstI, PvuII, RsaI, SacI, XbaI, XmnI on a panel of 8 unrelated Caucasians.

Chromosomal Location: 17q21-q22 based on location of HOX2A (2). Linkage data show no crossovers with the SacI RFLP identified by clone pBS3 (3).

Probe Availability: The pH2-1-9 clone is being sent to ATCC.

Acknowledgements: This work was supported by NIH grants MH39239 and GM40883 to KKK.

References: 1) Acampora, D. *et al.* (1989) *Nucl. Acids Res.* **17**, 10385-10402. 2) Solomon, E. and Barker, D. (1989) *HGM10: Cytogenet. Cell Gen.* **51**, 319-337. 3) Murphy, P.D. *et al.* (1987) *Nucl. Acids Res.* **15**, 6311.