A *Bam*HI RFLP at *MT2A* on human chromosome 16

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SOURCE/DESCRIPTION

Probe hMT-IIASC: a 770bp *Hind*III fragment from the 5' flanking region of *MT2A* subcloned into pUC13 (Karin and Richards, 1982).

POLYMORPHISM

*Bam*HI (New England Biolabs) identified one faint invariant band at 14.4kb and a simple 2 allele polymorphism with variant bands at 7.4kb and 2.9kb, in addition to the two *Taq*I polymorphisms previously described (Hyland et al., 1987).

FREQUENCY - From 35 unrelated Caucasians :

2.9kb allele (C1)	0.94
7.4kb allele (C2)	0.06

NOT POLYMORPHIC FOR

*Ban*I, *Bst*NI, *Eco*RV and others as previously described (Hyland et al., 1987), with a panel of 6 unrelated individuals.

CHROMOSOMAL LOCALISATION

16q21 by *in situ* hybridization to chromosomes expressing *FRA16B* and *FRA16C* (Simmers et al., 1987).

MENDELIAN INHERITANCE

Codominant inheritance was shown in Utah Family 984. Each parent was heterozygous for the 7.4kb and 2.9kb alleles with 4 offspring homozygous for the 7.4kb allele, 1 offspring heterozygous for each allele and 1 offspring homozygous for the 2.9kb allele.

PROBE AVAILABILITY

Available from R.I. Richards, Howard Florey Institute of Experimental Physiology and Medicine, University of Melbourne, Parkville, VIC. 3052

OTHER COMMENTS

No problems encountered with an overnight digestion using 4 units *Bam*HI/microgram genomic DNA and the recommended restriction buffer. Digests were carried out using 5mM spermidine to ensure complete digestion. One of the *Taq*I polymorphisms appeared to be in linkage disequilibrium with the *Bam*HI polymorphism. Allele C2 was associated with allele B2 in the 4 individuals observed with the C2 allele.

REFERENCES

1. Hyland V.J., Grist S., West A., Richards R.I. and Sutherland G.R. (1987): Nucl. Acids Res. 15:1350. 2. Karin M. and Richards R.I. (1982): Nature 299:797-802. 3. Simmers R.N., Sutherland G.R., West A. and Richards R.I. (1987): Science 236:92-94.

ACKNOWLEDGEMENTS

The Adelaide Children's Hospital Research Trust provided support.