An MboI polymorphism at codon 192 of the human tyrosinase gene is present in Asians and Afrocaribbeans

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Albinism is an inherited, generalised hypomelanotic condition, caused by impaired melanin synthesis (1). Tyrosinase catalyses the conversion of tyrosine into melanin. Mutations in its gene (*tyr*) have been described in a few albinos (2). A polymorphism for MboI at codon 192 of the human tyrosinase gene has been described in Caucasians but was absent from Indian Asians and Orientals (3,4). Hence, the region of tyrosinase encoded at the polymorphic site might determine the enzyme activity as ethnic pigmentation differences are due to the amount of melanin per cell and not the number of melanocytes (1).

PCR amplification used two primers 5'-GCTCCTGGC-TGTTTTGTACT-3' and 5'-CTGCCAGAGGAAGAATG-3' of exon 1 (5) 0.1−0.5μg genomic DNA, 1U Taq polymerase, 10pmol of each primer in 10μM dNTPs/15mM Tris-HCl pH 8.8/60mM KCl/2.25mM $MgCl_2$ for 30 cycles (92°C 15 sec, 53°C 5 sec, 72°C 40 sec). MboI digestion yields fragments of sizes 483, 247 and 87 bp (M1) when the segment contains two MboI sites (at codons 163 and 192). The absence of the MBOI 192 site yields bands of sizes 483 and 334 bp (M2).

The distribution of alleles in the Caucasians is similar to that reported (3) but the MboI 192 polymorphism is present in Indian Asians contrary to that report. It is also present in Afrocaribbeans. This has not previously been reported. We found no polymorphism in Orientals as in previous reports (3, 4). Its absence may be related to their separate development.

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Table 1. Distribution of alleles

	Alleles			
	M1/M1	M1/M2	M2/M2	Total
Caucasians	24	29	1	54
Indian Asians	8	17	1	26
Afrocaribbeans	12	6	2	20
Orientals	15	0	0	15
Total	59	52	4	115

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PCR detection of the insertion/deletion polymorphism of the human angiotensin converting enzyme gene (DCP1) (dipeptidyl carboxypeptidase 1)

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The insertion/deletion DNA polymorphism detected by the human angiotensin converting enzyme (DCP1) cDNA probe has been shown to be associated with the level of angiotensin converting enzyme (ACE) in the serum (1). The insertion was localized into intron 16 of the human DCP1 gene (2). DNA sequence of the insertion and of the flanking regions were determined by the chain termination method on a cloned genomic fragment of the DCP1 gene, pHA4.3 (2) (EMBL accession number X62855). The insertion itself corresponds to an *alu* repetitive sequence and is 287 bp long.

PCR Conditions: Reactions were performed with 10 pmoles of each primer: sense oligo 5' CTGGAGACCACTCCCATCCTTT-CT 3' and anti-sense oligo: 5' GATGTGGCCATCACATTCGT-CAGAT 3' in a final volume of 50 μl, containing 3 mM $MgCl_2$, 50 mM KCl, 10 mM Tris−HCl pH 8.4, 0.1 mg/ml gelatin, 0.5 mM of each dNTP (Pharmacia), 1 unit of *Taq* polymerase (Cetus) (3). The DNA was amplified for 30 cycles with denaturation at 94°C for 1 min, annealing at 58°C for 1 min, and extension at 72°C for 2 min using a PTC-100 thermal cycler (MJ Research). The PCR product is a 190 bp fragment in the absence of the insertion and a 490 bp fragment in the presence of the insertion. A third fragment with an intermediate molecular weight is present in PCR from heterozygotes, and likely corresponds to a heteroduplex DNA fragment.

Frequency/Inheritance: Codominant segregation of the insertion/deletion polymorphism was observed in 50 nuclear families. Allele frequencies were calculated from 199 unrelated individuals:
Deletion allele: 0.573
Insertion allele: 0.427

Chromosomal Location: The ACE gene has been assigned to 17q23 (4).

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