

MspI polymorphism of the human CYP2E gene

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Source/Description: Oligonucleotide primers were used to amplify a 480 bp segment of cytochrome P450IIE1 (CYP2E) intron VI (1) (GenBank accession no. J02843).

PCR Primers:

5' GTGCTGAACACTGGTACTCT 3'
5' CTGACTGGCCTGTTTGCATG 3'

Polymorphism: MspI detects a two-allele polymorphism with bands at 0.47 kb or 0.3 kb and 0.2 kb.

Frequency: Studied in 36 unrelated Japanese

| | | |
|-----------|---------------------|------|
| allele G1 | (0.47 kb) | 0.89 |
| allele G2 | (0.3 kb and 0.2 kb) | 0.11 |

Frequency of Heterozygosity: 0.22.

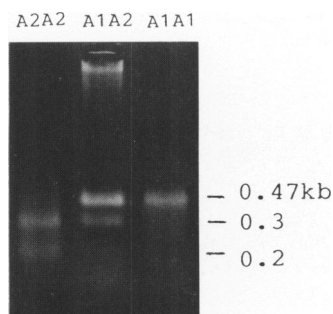
Chromosomal Localization: 10 (1).

Mendelian Inheritance: Autosomal co-dominant shown in one informative family.

Other Comments: This polymorphism was first detected using the cDNA probe λ hPD4 (2, 3). The PCR reaction was performed on 2 μ g of genomic DNA using 100 pmoles of each primer and 2.5 units of Taq polymerase. Samples were cycled thirty times at 94°C for 1 min, 50°C for 2 min and 72°C for 3 min.

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Dinucleotide repeat polymorphism at the human gene for insulin-like growth factor I (IGFI)

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Source/Description: The polymorphic (CT)_n repeat begins at base pair 140 of the human gene for insulin-like growth factor I on chromosome 12q23 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 187 bp.

Primer Sequences:

TTGTGTCAACTGCTGATATG (CT strand)
AACCAAAACATCATCCCTA (GA strand)

Frequency: Estimated from 78 chromosomes of unrelated individuals. Heterozygosity Index = 53%. PIC = 0.52.

| Allele (bp) | Frequency | Allele (bp) | Frequency |
|-------------|-----------|-------------|-----------|
| F1 207 | 0.01 | F7 195 | 0.01 |
| F2 205 | 0.03 | F8 193 | 0.05 |
| F3 203 | 0.03 | F9 191 | 0.03 |
| F4 201 | 0.03 | F10 189 | 0.67 |
| F5 199 | 0.10 | F11 173 | 0.01 |
| F6 197 | 0.03 | | |

Mendelian Inheritance: Co-dominant segregation was observed in two informative families.

Chromosomal Localization: The human gene for insulin-like growth factor I has been assigned to chromosome 12q23 (3).

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described (4) except that the denaturation cycle at 94°C was extended to 1.4 minutes. The dinucleotide repeat was based on a (CT)₁₆ sequence.

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