TaqI RFLP of the human tropomyosin gene (TPM3) involved in the generation of the TRK oncogene

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Source/Description: The 1.0 Kb HindIII-PvuII fragment of plasmid pDM-8 (A1), related to a non-muscle protein isoform of tropomyosin gene found to form part of the TRK oncogene (2), was used as a hybridization probe.

Polymorphism: TaqI digestion identifies a biallelic RFLP with bands of 3.9 (1) and 3.7 (A2) kb.

Frequency: Analyzed in 87 unrelated Italian Caucasians and in 77 unrelated members of CEPH families.

Allele	Frequency
A1	0.33
A2	0.67

Not Polymorphic For: HindIII, BamHI, BglII (each enzyme tested on at least 14 unrelated individuals).

Chromosomal Localization: TPM3 has been assigned to 1q31 both by hybridization to a panel of somatic cells hybrids and by 'in situ' hybridization (P. Radice *et al.*, (1991) *Oncogene*, in press).

Mendelian Inheritance: Co-dominant segregation has been shown in four two-generation families.

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The MspI polymorphism in intron 6 of p53 (TP53) detected by digestion of PCR products

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Source/Description: A 107 bp fragment of intron 6 of the TP53 gene was amplified from human genomic DNA using the polymerase chain reaction (PCR). A polymorphism was identified by sequencing intron 6. GenBank/EMBL accession no. X54156.

Polymorphism: MspI identifies a 2-allele polymorphism: (E1: 63 + 44 bp; E2: 107 bp).

Allelic Frequency: Estimated in 57 unrelated Americans: A1 = 0.74, A2 = 0.26. Observed heterozygosity = 46%.

Chromosomal Localization: The polymorphic MspI recognition site is located within the 6th intron of the human TP53 locus 60 bp downstream of the 3' end of exon 6, on chromosome 17p13.1 (1).

Mendelian Inheritance: Co-dominant segregation demonstrated in one 2 generation family with 6 members.

PCR Primers:

upstream: 5'-AGGTCTGGTTTGCAACTGGG-3'
downstream: 5'-GAGGTCAAATAAGCAGCAGG-3'

PCR Conditions: Reactions (100 μ l) consisted of: 50 ng genomic DNA, 20 pmol each primer, 2 units Promega Taq DNA polymerase (Promega, Madison, WI), 1 $\times$ Promega Taq polymerase buffer, 75 μ mol each dNTP. Amplification was carried out in an Ericomp Programmable Cyclic Reaction (Ericomp, San Diego, CA) as follows: 1 cycle of: 10 min at 95°C, 2 min at 59°C, 1 min at 72°C; followed by 35 cycles of: 1 min at 95°C, 1 min at 59°C, 1 min at 72°C.

Other Comments: this polymorphism may be identical to a p53 MspI polymorphism that de la Calle-Martín *et al.* characterized using Southern blots (2); our allelic frequencies are similar to those published by this group. The precise location of this previous polymorphism has not been reported.

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