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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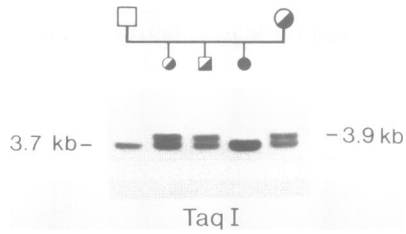

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The MspI polymorphism in intron 6 of *p53* (TP53) detected by digestion of PCR products

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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: *Msp*I 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 *Msp*I 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 μl) consisted of: 50 ng genomic DNA, 20 pmol each primer, 2 units Promega *Taq* DNA polymerase (Promega, Madison, WI), 1×Promega *Taq* polymerase buffer, 75 μmol 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* *Msp*I 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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