A novel BamHI polymorphism for the human transforming growth factor alpha gene (TGF-α)

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Source/Description: pJF-17 contains 390 bp of exon VI and 66 bp of intron V, cloned into the HindIII-EcoRI sites of pBS (+). pJF-26 contains 31 bp of 5'-UT, 480 bp of TGF-α precursor and 385 bp of 3'-UT sequences and pJF 25 contains 3.2 kb of 3'-UT sequences and a poly A tract, cloned into the EcoRI site of pBluescript KS (+). The plasmids pJF-25 and pJF-26 correspond to Murray's plasmids ph TGF1-10-3350 and ph TGF1-10-925, respectively (Murray *et al.* 1986).

Polymorphism: BamHI identifies two-allele polymorphisms with pJF-17, pJF-25 and pJF-26.

Frequency: Studied in 73 Caucasians:
10 kb allele 0.062
7 kb allele 0.938

Not Polymorphic For: BclI, BglII, EcoRI, EcoRV, HaeIII, HindIII, HinfI, KpnI, MspI, PstI, PvuII, SacI, XbaI, XhoI (Murray *et al.* 1986).

Chromosomal Localization: 2p13 (Tricoli *et al.*, 1985).

Mendelian Inheritance: Demonstrated in two families.

Probe Availability: Contact J.F.Qian.

Other Comments: The two-allele RFLPs of TaqI and RsaI described by Murray *et al.* (1986), were detected with pJF-17, pJF-25 and pJF-26. With pJF-25 and pJF-26, constant bands were detected in addition to the two alleles RFLPs; this was not observed for the pJF-17. Murray *et al.* already described a BamHI polymorphism, however, their data in terms of fragment lengths and relative frequencies, were somewhat different (7.0 kb 0.17; 4.0 kb 0.83).

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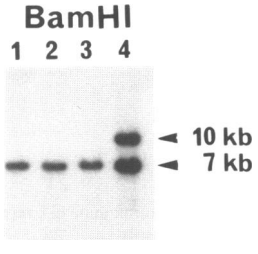
References: 1) Tricoli,J.V. *et al.* (1985) *Cyto. and Cell Genet.* **40**, 762. 2) Murray,J.C. *et al.* (1986) *Nucl. Acids Res.* **14**, 7136.



AT repeat polymorphism at the D5S122 locus tightly linked to adenomatous polyposis coli (APC)

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Source and Description: The polymorphic AT repeat AT83a was isolated and characterized from cosmid cCB83 (D5S122). Cosmid cCB83 was derived from a reduced complexity radiation hybrid containing fragments of human chromosomes 4 and 5 in a Chinese hamster background (1). From the same cosmid an MspI RFLP was previously characterized (2).

Primer Sequence:
AT primer = CB83a.AT 5'-GTAGGGGTAAATTAGCCTCTC-3'
TA primer = CB83a.TA 5'-GATAATGGAGGAGGCTATGC-3'

Polymorphism and Frequency: Two alleles were detected in 57 unrelated Dutch volunteers.

Allele	Number of AT repeats	Frequency	Product size (bp)
B1	8	0.14	213
B2	6	0.86	211

Haplotype frequency of the AT repeat polymorphism and the MspI RFLP (2):
A1/B1 = 0.025, A1/B2 = 0.000, A2/B1 = 0.300, A2/B2 = 0.100, A3/B1 = 0.550, A3/B2 = 0.025
PIC = 0.19
Combined PIC = 0.54

Mendelian Inheritance: Autosomal co-dominant segregation was observed in 25 Dutch families.

Clinical Relevance: Presymptomatic diagnosis of familial adenomatous polyposis (FAP). In 39 informative phase known meioses in 6 Dutch families with the recombinants, $\theta = 0.00$, $Z = 8.63$ between D5S122 and APC.

Chromosomal Localization: Assigned to the long arm of chromosome 5 (q15-q23) by using a panel of human-Chinese hamster radiation hybrids and by fluorescent in situ hybridization (1).

Other Comments: The products of the PCR reactions (performed according to standard procedure) were loaded on 10% denaturing (8 M urea) polyacrylamide gels in TAE buffer. The electrophoresis is performed overnight at 12.5 V/cm in an 'aquarium tank' filed with buffer maintained at 60°C. The gel is then stained with ethidium bromide and the bands are directly observed under U.V. light.

References: 1) Tops,C.M.J. *et al.* in preparation. 2) Breukel *et al.* (1991) *Nucl. Acids Res.* **19**, 685.

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