Taql RFLP at D21S137
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Source and Description: D32 is a probe from human chromosome 21 that was isolated using Alu-mediated recombination (1). It is a 4.4 kb EcoRI/HindIII fragment subcloned into pIA7, which was subcloned into pMLC28 (2) by us.

Polymorphisms: Taql identifies a two allele polymorphism with fragment sizes of 4.2 (allele A2) and 6.3 kb (allele A1), with an additional invariant band of 0.6 kb.

Frequency: Studied in 13 unrelated European Caucasians.
allele 1: 0.4
allele 2: 0.6

Chromosomal Location: D21S137 has been assigned to 21q11.2-q21 by somatic cell hybrids (1). We have found this probe to lie distal to D21S52 (3).

Mendelian Inheritance: Co-dominant inheritance observed in one large extended pedigree.

Probe Availability: Contact U. Tantravahi (Department of Obstetrics & Gynecology, Columbia University, New York).

Other Comments: High stringency hybridization and washing are required to see unique bands with this probe. Incomplete digestion with Taql can falsely mimic the presence of the larger allele.

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A Taql RFLP at the human TOP1 pseudogene locus on chromosome 22q11.2-13.1 (TOP1P2)
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Source and Description: A 3.6 kb cDNA clone, T1B, obtained by screening a human λgt11 library (1), was subcloned into the EcoRI site of Bluescript.

Polymorphism: After Taql digestion of genomic DNA the probe detects three allelic fragments of 8.4, 7.5 and 5.2 kb. Constant bands are 6.5, 4.9, 3.9, 3.3, 2.8, 2.1, 1.6, 1.4 (2 closely spaced bands) and 1.0 kb.

Frequency: Estimated from 70 unrelated Caucasians:
8.4 kb allele (A1) 0.34;
7.5 kb allele (A2) 0.59;
5.2 kb allele (A3) 0.07. Observed heterozygosity: 56%.

Not Polymorphic For: PstI, EcoRI, Rsal, MspI, HindIII and BglII (studied in at least 15 unrelated individuals).

Chromosomal Localization: The 3 polymorphic bands were mapped to chromosome 22 using a somatic cell hybrid panel. The 2.1 kb constant band mapped to chromosome 1q whereas all other constant bands mapped to chromosome 20. Whereas the active gene for hTOPl is located on chromosome 20q11.2–13.1, two pseudogenes are on chromosomes 1q23–24 and 22q11.2–13.1 (2). The sequences of hTOPl and the two pseudogenes have been determined (1, 3).

Mendelian Inheritance: Co-dominant segregation demonstrated in a three generation family (24 meioses).

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