

TaqI RFLPs at the Wilms' tumor gene (WT1)

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Source/Description: Clone WT33 contains a 2.3 kb cDNA of one of the candidate genes for Wilms' tumor at 11p13 (1). A 1.8 kb EcoRI fragment of the plasmid was used as a probe, which covered the 5' portion of the gene.

Polymorphisms: TaqI detected two 2-allele polymorphisms (set Aa and Ab) with fragment size of 3.4 kb (A1) or 3.2 kb (A2), and 2.2 kb (A1) or 1.5 kb (A2). Constant bands of 6.6, 3.7, 1.7, 1.2 and 0.8 kb were also detected.

Frequencies: Studied in 65 unrelated individuals of the normal Japanese (2). Frequencies of haplotype were estimated with the aid of tumor DNAs with loss of heterozygosity.

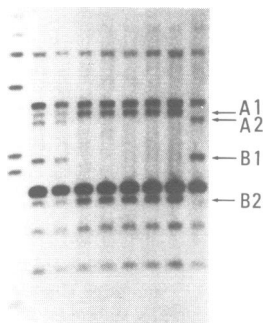
| Set | Alleles | Set | Alleles | |
|-----|----------|-----|----------|-------------|
| Aa | A1 = .88 | Ab | A1 = .14 | a1b1 = .02 |
| | A2 = .12 | | A2 = .86 | a1b2 = .86 |
| | | | | a2 b1 = .12 |
| | | | | a2 b2 = .00 |

Mendelian Inheritance: Co-dominant segregation was confirmed in 15 two-generation families.

Other Comments: The above two polymorphisms seemed to be in linkage disequilibrium. The polymorphism was also observed in the Wilms' patients with the allelic frequencies being similar. Therefore, a particular allele seemed not to associate with genesis of the tumor. Nevertheless, this polymorphism was useful to detect loss of allele at the locus.

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MspI RFLP detected by a ZNF-40 cDNA sequence

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Source/Description: 1.6 kb EcoRI fragment from a 5' cDNA clone (designated 4b) of the human ZNF-40 locus. ZNF-40 (MBP-1) (PRDII-BF1) encodes the class I major histocompatibility complex (MHC) enhancer binding protein 1 (2).

Polymorphism: MspI digestion of genomic DNA and hybridization with this probe reveals a two-allele polymorphism with fragment sizes of 1.60 kb (A1) and 1.85 kb (A2). Two additional faint constant bands of 1.90 kb and 0.5 kb are also detected by this probe.

Frequency: Allele frequencies were determined from 94 unrelated individuals, including 79 CEPH parent DNAs. A1 = 0.26 A2 = 0.74

Not Polymorphic For: BanII and HindIII (≥ 4 individuals screened).

Chromosomal Location: Initial localization to chromosome 6 was determined using somatic cell hybrids, followed by more precise assignment to 6p23-24 using *in situ* and multipoint mapping techniques (3).

Mendelian Inheritance: Co-dominant segregation was observed in 21 informative CEPH families (248 individuals) (3).

Probe Availability: Request for probe to Dr.A.S.Baldwin at the above address.

Other Comments: This RFLP was observed under normal hybridization and washing (i.e. 0.1 \times SSC 0.1% SDS at 65°C for 15 minutes) conditions. A reduction in stringency is not recommended when using this probe due to the presence of a related gene/pseudogene in the genome (2).

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