A second useful polymorphism for the cytosolic thymidine kinase gene (TK1) with the enzyme BstEII which will allow haplotyping at this locus on chromosome 17 (q21-q22)

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SOURCE/DESCRIPTION: pHtk9 is a cDNA clone (Lin et al., 1985) of human cytosolic TK isolated from the cDNA library of Okayama and Berg (Okayama and Berg, 1983).

POLYMORPHISM: BstEII (G/GTNACC) (New England Biolabs) detects a codominant three allele system with bands at 2.1 kb or [1.0 kb and 1.1 kb], or [1.0 kb and 0.6 kb].

FREQUENCY: Studied 43 unrelated European Caucasians:

2.1 kb allele (B1) : 0.34 ± .01
1.1 kb and 1.0 kb (B2) : 0.02 ± .01
1.0 kb and 0.6 kb (B3) : 0.64 ± .01


CHROMOSOMAL LOCALISATION: 17q21-q22 (Kit, 1976; Willecke et al., 1977).

MENDELIAN INHERITANCE: Autosomal codominant segregation observed in four large reference pedigrees with 14 informative matings.

PROBE AVAILABILITY: F.H. Ruddle, Department of Biology, Yale University, New Haven, CT 06510 USA.

OTHER COMMENTS: A frequent TaqI polymorphism and rare HindIII and PstI polymorphisms have been previously reported (Murphy et al., 1986)

REFERENCES:

ACKNOWLEDGEMENTS: Funded by USPHS grants CA32066 and MH39239 (K. Kidd) and GM09966 (F. Ruddle).