A frequent polymorphism for the cytosolic thymidine kinase gene, TK1, (17q21-q22) detected by the enzyme TaqI

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SOURCE/DESCRIPTION: pHtk9, a 1.4 kb cDNA clone of the human cytosolic thymidine kinase gene (Lin, unpublished observations) inserted into pcD (Okayama and Berg, 1983).

POLYMORPHISM: TaqI (T/CGA) (New England Biolabs) identifies one nonvariant band at 1.6 kb and a simple two allele polymorphism with a band at either 4.3 kb or 1.3 kb.

FREQUENCY: Studied 25 European Caucasians.
4.3 kb allele (A1) .60 ± .01
1.3 kb allele (A2) .40 ± .01

NOT POLYMORPHIC FOR: ApaI, BamHI, BglII, BstNI, CfoI, EcoRI, EcoRV, HaeIII, HincII, HinfI, HpaII, KpnI, MspI, PvuII, RsaI, SacI, ScaI XbaI, XhoI, XmnI, all with a panel of 1-012 unrelated Caucasians.

CHROMOSOMAL LOCALISTION: 17q21-q22 (Kucherlapati et al., 1974).

MENDELIAN INHERITANCE: Autusomal codominant inheritance is seen in three kindreds with a total of 125 individuals.

PROBE AVAILABILITY: Available for collaboration; contract P.F. Lin at Bristol-Yale Laboratory, Yale University, Sterling Hall of Medicine, Room C-10, New Haven, CT 06510.

OTHER COMMENTS: Rare variants were seen with HindIII and PstI but have not yet been fully characterised.

REFERENCE: Okayama, H. and Berg, P. Molc. Cell Biol. 3: 280-289 (1983).
Kucherlapati, R., McCougall, JK and Ruddle, F,H. Cytogenetic. Cell Genet. 13: 108-110 (1974).

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