Human T-cell receptor CD3- ϵ (CD3E) / TaqI DNA polymorphism

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SOURCE/DESCRIPTION: 1.5 kb cDNA of the CD3-epsilon subunit of the human CD3 (T3) antigen/T-cell antigen receptor complex (1).

<u>POLYMORPHISM</u>: TaqI (TCGA) detects a single two-allele polymorphism with bands at either 8.7 kb or 8.1 kb.

FREQUENCY: Estimated from 107 unrelated Caucasians.

TaqI 8.7 kb allele: 0.67 8.1 kb allele: 0.33

NOT POLYMORPHIC FOR: BamHI, BglII, EcoRV, MspI and PvuII using a panel of 12 unrelated individuals.

<u>CHROMOSOMAL LOCATION</u>: Assigned to human chromosome 11q23 by Southern blot analysis of human-rodent somatic cell hybrids, and by *in situ* hybridization (2).

<u>MENDELIAN INHERITANCE</u>: Co-dominant segregation of the TaqI alleles observed in 31 families (383 individuals). Genotypes occur in Hardy-Weinberg proportions.

PROBE AVAILABILITY: Request for probe to C. Terhorst at the
above address.

OTHER COMMENTS: RFLPs were observed under normal hybridization and washing conditions.

REFERENCES: 1. D.P. Gold et al., Nature 321:431-434 (1986)
2. D.P. Gold et al., Proc. Natl. Acad. Sci. 84:1664-1668 (1987)

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