Human T-cell receptor CD3-6 (CD3D) / MspI DNA polymorphism

Uma Malhotra and Patrick Concannon

Virginia Mason Research Center, 1000 Seneca Street, Seattle, WA 98101, USA

SOURCE/DESCRIPTION: Plasmid pPGBC9 (1) containing a complete cDNA clone of the human CD3-delta gene was cleaved with BamHI. Fragments of 0.5 kb and 0.6 kb were isolated, pooled and used as a probe.

<u>POLYMORPHISM</u>: MspI (CCGG) detects allelic fragments 5.4 kb and 4.6 kb.

FREQUENCY: Estimated from 84 unrelated individuals.

5.4 kb allele: 0.06 4.6 kb allele: 0.94

<u>NOT POLYMORPHIC FOR</u>: RsaI, BstEII, EcoRV, BamHI, EcoRV, HindIII, BglII, PvuII, BglI, and PstI in at least 6 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>: Codominant segregation demonstrated in 4 unrelated families (49 individuals).

PROBE AVAILABILITY: Available from the American Type Culture Collection, 12301 Parklawn Drive, Rockville MD 20852.

<u>OTHER COMMENTS</u>: This probe also detects a TaqI RFLP with allelic fragments of 2.0 kb and 1.8 kb. TaqI and MspI alleles are in strong linkage disequilibrium.

REFERENCES: 1. van den Elsen, P., et al., Nature 312:413-18
(1984)
2. Gold, D.P., et al., Proc. Natl. Acad. Sci. USA 84:1664-1668
(1987)

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