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Rare genetic variant register

A rare genetic variant of the T cell receptor gamma joining segment TRGJI

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Summary. A rare *Hin*dIII restriction fragment variant of the human T cell receptor gamma joining segment TRGJ1 has been identified, in addition to two previously described alleles.

Source/description. The TRG1.4 probe (Bensmana et al. 1991) is a genomic 1450-bp *Eco*RI-*Hin*dIII fragment cloned in pUC18, and located between the TRGJ1 joining segment and the exon 1 of TRGC1 (Lefranc and Rabbitts 1985; Lefranc et al. 1986). This probe cross-hybridizes to the corresponding region located between TRGJ2 and TRGC2.

*Polymorphism. Hin*dIII (A/AGCTT) identifies three allelic restriction fragments of 2.1 kb *(*A1)*, 5.0 kb *(*A2)* and 3.7 kb *(*A3)*. Constant band: 2.1 kb.

*Frequency. *A1:* 0.64, **A2:* 0.35, **A3:* 0.01, as studied in 52 unrelated Caucasoids; the variant was not polymorphic for *Eco*RI (5 kb + 7 kb), *Bam*HI (20 kb + 13 kb), *Kpn*I (9 kb + 16 kb), *Sac*I (6.4 kb + 7.0 kb) studied in 25 unrelated individuals. The two constant bands observed with these four enzymes correspond to the TRGC1 and TRGC2 regions, respectively.

Chromosomal localisation. The human TRG locus has been mapped on chromosome 7 (Rabbitts et al. 1985) at band p14–p15 (Murre et al. 1985; Bensmana et al. 1991).

Mendelian Inheritance. Co-dominant segregation was demonstrated in five families.

Probe availibility. Available from Dr. M.-P. Lefranc.

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Other comments. The two alleles, **A1* (2.1 kb) and **A2* (5.0 kb), have been previously described using the TRGJ1 probe, pH60 (Lefranc and Rabbitts 1985; Lefranc et al. 1986; Buresi et al. 1989). In this report, we describe a third allele, **A3* (3.7 kb), corresponding to the absence of the *Hin*dIII site located 160 bp upstream of TRGJ1. The location of this polymorphic *Hin*dIII site 5′ of TRGJ1 does not interfere with the sizes of the Vγ-Jγ rearranged bands and therefore with the assignment of the given Vγ genes and Jγ segments, as described by Forster et al. (1987).

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