HindIII restriction fragment alleles of the human immunoglobulin \( \lambda \)II subgroup genes

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Source/Description: The VXII probe, clone pVX2EK0.3 is a 310 bp EcoRI-KpnI genomic fragment of the VX2.1 (IGLV2S1) gene isolated from the \( \lambda 275 \lambda 2 \) clone (1) subcloned in pUC19.  
Polymorphism: HindIII (A/AGCTT) identifies two allelic restriction fragments of 16.0 kb (IGLV2S*A1), 9.0 kb (IGLV2S*A2).  
Frequency: Studied in 34 unrelated Caucasoids.  
IGLV2S*A1: 50%, IGLV2S*A2: 50%  
Chromosomal Localisation: 22q11.  
Mendelian Inheritance: Co-dominant segregation, studied in one family.  
Probe Availability: Dr M.-P.Lefranc.

Other Comments: Run on 0.8% agarose gels and hybridized under high stringency conditions. ‘IGLV2S’ stands for ‘human immunoglobulin lambda variable gene belonging to subgroup II’. ‘IGLV2S1’ stands for \( \lambda 2.1 \) following the Human Gene Mapping recommendation (HGM9). The same polymorphism is detected at low stringency by cross-hybridization by the VXIIIII probe, clone pVX3RC0.15, a 144 bp EcoRI-BclI fragment isolated from the LY67C03-6 clone (2).


D21S170 maps to terminal 21q22.3

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Source/Description: F33B4 (D21S170) is a 4.9 kb BamHI fragment derived from a C2RB cosmid clone obtained by screening a library constructed from the CHO-human hybrid cell line 72532x-6.

Polymorphism: BamHI detects a three allele polymorphism with bands at 5.2 kb (A1), 4.9 kb (A2), and 4.8 kb (A3). Several constant bands are also detected.

Frequency: Estimated from 33 unrelated Caucasians:  
A1 = 0.24  
A2 = 0.71  
A3 = 0.05

Not Polymorphic For: PstI, HindIII, BglIII.

Chromosomal Localization: The probe was mapped to 21q22.3 by using a somatic cell hybrid panel. Refined mapping studies based on the use of hybrids containing different portions of this human DNA segment (1) have allowed assignment of D21S170 to the terminal part of q22.3.

Mendelian Inheritance: Co-dominant segregation was observed in 5 families including one family with a Down syndrome child with a de novo 14/21 translocation which most likely originated during maternal meiosis (2). The probe detected 2 doses of the A2 allele and one dose of the A1 allele in the child showing its usefulness in the study of Down syndrome families (see Figure).

Probe Availability: Contact C.Brahe.

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Allelic bands described in this report are shown by triangles.

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