Repeat polymorphisms in the interleukin-4 gene (IL4)

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Source/Description: Several mini repeat sequences in hematopoietic growth factors genes localized on chromosome 5 were analyzed for polymorphisms by polymerase chain reaction (PCR). For interleukin-4 (IL-4) (1), two polymorphic markers were identified: a GT dinucleotide repeat in intron 3 (A), and a 70 bp repeat in intron 2 (B).

PCR Primers:

IL-4 RP1: A 5'-CTCAAAGTGCTGGGATTAGC-3'
 B 5'-AGCCATCTCGTTGGATGGA-3'
 IL-4 RP2: A 5'-TAGGCTGAAAGGGGAAAGC-3'
 B 5'-CTGTTACCTCAACTGCTCC-3'

Polymorphism and Frequency: Estimated from 33 unrelated individuals:

IL-4 RP1:		IL-4 RP2:	
Allele (bp)	Frequency	Allele (bp)	Frequency
A1: 184	0.73	B1: 183	0.17
A2: 196	0.07	B2: 253	0.83
A3: 206	0.20	heterozygosity: 0.12	
heterozygosity: 0.27			

Chromosomal Localization: IL-4 is located on chromosome 5q23.3–31.2 (2).

Other Comments: PCR was carried out in a volume of 50 μ l containing 50 ng genomic DNA, 50 pmol of each set of primers (A and B), 250 μ M of each dNTP's, 50 mM KCl, 10 mM Tris-HCl pH 8.3, 2.0 mM MgCl₂, 0.2 mg/ml nuclease free BSA, 1 unit Taq polymerase (Cetus AmpliTaq). The amplification consisted of 27 cycles of 0.5 min at 95°C, 0.7 min at 55°C and 0.7 min at 72°C. Allelic fragments were resolved on either 3% agarose gels or on a 6% acrylamide gel with or without 10% glycerol as described (3) and stained with ethidium bromide.

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A KpnI RFLP at the human myeloperoxidase locus

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Source/Description: A myeloperoxidase cDNA clone (pMP23: 3.05 Kb EcoRI insert in pBR322) lacking 220 bases on the 5' end was used as probe (1).

Polymorphism: KpnI detects a two-allele polymorphism: 7.5 Kb (A1) or 5.5 Kb/2 Kb (A2). Constant bands of 20 and 6 Kb are also seen.

Frequency: Estimated from 75 unrelated individuals:

C1: 0.13

C2: 0.87

Not Polymorphic For: EcoRI, BamHI, HindIII, BglII.

Chromosomal Localization: Localizations reported in the literature include:

17q21-q23 (2, 3).

Mendelian Inheritance: Autosomal codominant segregation was observed in 2 families.

Probe Availability: Obtained through ATCC.

Other Comments: A PstI RFLP (4) and an EcoRV (5) RFLP have been previously reported. There is no linkage disequilibrium with the EcoRV RFLP.

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