HindIII RFLP at the human IFNAR locus

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Source/Description: A 195 bp EcoRI-SalI fragment 19ES was used as an hybridization probe. It derives from a cDNA for the gene IFNAR which encodes the human interferon alpha receptor (1) (GenBank Accession Number J03171), and contains upstream non-coding and coding sequences (amino-acids 1–39).

Polymorphism: HindIII identifies a 5-allele polymorphism with bands at 11.8 kb (A1), 10.9 kb (A2), 9.3 kb (A3), 8.9 kb (A4), 8.1 kb (A5).

Frequency: Estimated from 31 unrelated individuals:

A1: 0.23

A2: 0.35

A3: 0.24 PIC = 0.71

A4: 0.10

A5: 0.08

Not Polymorphic For: MspI, PstI.

Chromosomal Localization: The human IFNAR gene maps to 21q22.1 (2).

Mendelian Inheritance: Co-dominant segregation of the HindIII RFLP was observed in a three-generation family in which 17 individuals were typed (11 informative meioses).

Probe Availability: The probe will be submitted to the American Tissue Culture Collection (ATCC). Until it is available through the ATCC, it can be obtained from E.V.

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A PstI polymorphism at the D5S39 locus

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Source/Description: Probe p105–153 Ra (D5S39) is a 4 kb fragment cloned into the EcoRI site of PUC8, isolated from a human genomic library (1).

Polymorphism: PstI identifies a two-allele polymorphism (C₁: 3.5; C₂: 3.2) in addition to the two-allele polymorphisms described with MspI and XbaI (2).

Frequency: The allele frequencies were estimated from 50 unrelated individuals.

C₁: 0.20

C₂: 0.80

Not Polymorphic For: PvuII, TaqI, BglII, RsaI.

Chromosomal Localization: This probe has been assigned to 5q12-q14 (using linkage analysis) (1–2).

Mendelian Inheritance: Co-dominant segregation demonstrated in 5 families.

Availability: ATCC.

Other Comments: This probe has recently been shown to be tightly linked to the SMA locus (3). The use of the three enzymes MspI, XbaI, PstI increases the number of informative meioses.

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