Two RFLPs in human inter-alpha-trypsin inhibitor heavy chain gene ITIH2 on chromosome 10

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SOURCE AND DESCRIPTION OF CLONE: The 0.8 kb Eco RI / Bam HI fragment of lambda HuHITI-9 (1) used as a probe codes for human heavy chain H2 of the inter-alpha-trypsin inhibitor (2).

POLYMORPHISMS:
Kpn I (GGTAC/C) identifies one invariant band at 8.5 kb and a diallelic polymorphism with DNA fragments at 26.0 kb or 20.0 kb.

Msp I (C/CGG) identifies three invariant bands at 2.35 kb, 2.1 kb and 1.0 kb and a diallelic polymorphism with DNA fragments at 5.0 kb or 2.8 kb and 2.2 kb.

FREQUENCIES:
Kpn I:
- Studied in 30 healthy Caucasians.
- 20.0 kb allele (0.30)
- 26.0 kb allele (0.70)

Msp I:
- Studied in 16 healthy Caucasians.
- 5.0 kb allele (0.19)
- 2.8 kb and 2.2 kb allele (0.81)

NOT POLYMORPHIC FOR: Apa I, Ava II, Bam HI, Bcl I, Bst EII, Eco O109, Eco RI, Eco RV, Hind III, Hinf I, Nhe I, Sst I, Sca I and Xba I.

CHROMOSOMAL LOCALISATION: The ITIH2 gene has been mapped to 10p15 by in situ hybridization (2).

MENDELIAN INHERITANCE: Codominant segregation (fig.) was found for each polymorphism in two informative families.

PROBE AVAILABILITY: Available from J. P. Martin.

REFERENCES: