RsaI polymorphism of a human immunoglobulin VH5 subclass locus

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SOURCE AND DESCRIPTION OF CLONE: A 221 bp PstI fragment from clone 2-V specific for human VH5 subclass genes

POLYMORPHISM of a VH5 gene detected with RsaI (GT/AC). Presence or absence of a fragment of 3.2 kb (two fragments of 1.8 kb and 2.2 kb are constant).

FREQUENCY: Studied in DNA from 24 random Canadian caucasians:

Frequency of 3.2 kb fragment: 0.50

Frequency of the absence of the fragment: 0.50

NOT POLYMORPHIC FOR: ApaI, BglI, BstEII, HinfI, PvuII, TaqI, SacI, Sau3AI in digests of DNA from 7 unrelated individuals.

CHROMOSOMAL LOCALIZATION: In IGHV cluster at 14q32.3 to 14qter (1), confirmed by PFGE (3).

MENDELIAN INHERITANCE: Codominant pattern of inheritance shown in 6 three generation families (83 individuals).

PROBE AVAILABILITY: Write to Frederick W.Alt, Columbia University.

OTHER COMMENTS: Wash stringency of 0.1% SDS, 0.1x SSC; at 50°C. This specific VH5 fragment has been reported to be rearranged in two patients with chronic lymphocytic leukemia (2).

REFERENCES: (1) Cox D.W. et al, 1982; Nature 297, 428-430.
(2) Shen A. et al, 1987; P.N.A.S. USA 84, 8563-8567.
(3) Berman J.E. et al, (1988); Embo J7, 727-738

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