PstI RFLP in the human hexosaminidase (*HEXB*) gene on chromosome 5

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SOURCE/DESCRIPTION: The probe is a 1.0 kb PstI fragment obtained from a portion of the *HEXB* gene subcloned in pSP64. This fragment consists mostly of intron sequences, with ~100 bp of coding sequence. The subcloned fragment was obtained from a recombinant phage carrying part of the *HEXB* gene isolated from a library of human genomic DNA (1) by screening with a hexosaminidase beta-chain cDNA (2).

POLYMORPHISM: PstI (CTGCAG) detects a simple two-allele polymorphism, with a band at either 1.6 kb (A1) or 1.0 kb (A2). An invariant band of length 1.2 kb is also detected.

FREQUENCY: 216 chromosomes were scored from 108 unrelated Caucasian individuals.

1.6 kb allele (A1): 0.12
1.0 kb allele (A2): 0.88

NOT POLYMORPHIC FOR: The cDNA clone (2) is not polymorphic with EcoRI, PvuII, or HindIII. Other enzymes have not been studied extensively.

CHROMOSOMAL LOCALIZATION: Chromosome 5q13 (HGM8)

MENDELIAN INHERITANCE: Co-dominant segregation demonstrated in several large, three-generation families (C.E.P.H.).

PROBE AVAILABILITY: Generally available. Contact R.A.G. at Department of Genetics, Hospital for Sick Children, Toronto, Ontario M5G 1X8

OTHER COMMENTS: Low background with final wash in 0.1 x SSC at 65°C.

REFERENCES: (1) Lawn et al., Cell 15: 1157-1174 (1978)
(2) O'Dowd et al., PNAS 82: 1184-1188 (1985)

ACKNOWLEDGEMENTS: Supported by grant PG-4 from the Medical Research Council of Canada to H.F.W., R.A.G., and D.M.