Pvu II RFLP at the human chromosome 1 alpha-L-fucosidase gene locus (FUCA1)

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SOURCE AND DESCRIPTION OF CLONE: The fucosidase clone AF₃ was isolated from a lambda gt₁₁ human hepatoma cDNA library and subcloned in pUC 18 (Fukushima et al, 1985). AF₃ is a 1058 bp cDNA which encodes for approximately 80% of the mature alpha-L-fucosidase. The 619 bp probe used in this study is a 3' fragment of AF₃ obtained from a Hinc II digest.

POLYMORPHISM; Pvu II (CAG/CTG) (New England Biolabs) identifies two invariant bands at 3.0 kb and 4.5 kb and a simple two allele polymorphism with a band at either 7.0 kb (A) or 6.0 kb (B).

FREQUENCY: Studied 32 random unrelated Caucasians.

7.0 kb allele (A) 0.750

6.0 kb allele (B) 0.250

NOT POLYMORPHIC FOR: Ava II, Bam HI, Bgl II, Eco RI, Eco RV, Kpn I, Mbo I, Msp I, Pst I, Rsa I, Sac I, Stu I, Taq I, or Xmn I with DNA from 8 unrelated Caucasians.

CHROMOSOMAL LOCALISATION: The human alpha-L-fucosidase gene was assigned to chromosome 1p34.1 - 1p36.1 by somatic cell hybrids and by in-situ hybridization (Fowler et al, 1986). This RFLP was also mapped to the short arm of chromosome 1 by somatic cell hybrids. Sequences are also detected on chromosome 2 but without DNA polymorphism.

MENDELIAN INHERITANCE Co-dominant inheritance seen in 10 families (100 individuals.)

PROBE AVAILABILITY: Available for collaboration by request to J. S. O'Brien.

OTHER COMMENTS: Low background at wash stringency of .2 x SSPE at 60 degrees centigrade. Can be used as a DNA or RNA probe.

REFERENCES: Fukushima et al., "Molecular cloning of a cDNA for human alpha-L-fucosidase", PNAS 82 (1985) 1262-1265.

Fowler et al., "Chromosome 1 localization of the human alpha-L-fucosidase structural gene with a homologous site on chromosome 2", Cytogenet. Cell Genet. (1986) (v. 43:in press).

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