A HindIII RFLP at the FTHP1 locus on chromosome 6

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Source/Description: Probe FADJ3 is a 0.7 Kb fragment derived from the 3' flanking region of a H-ferritin pseudogene FTHP1 (previously FTHL5) mapped to 6p12-6p21.3 (1). FTHP1 was cloned as a 12 Kb HindIII fragment in lambda L47-1 clone and FADJ3 subcloned as a KpnI-EcoRV fragment in the Bluescript plasmid.

Polymorphism: HindIII identifies a biallelic polymorphism (12 Kb and 9 Kb).

Frequency: Studied in 65 unrelated caucasoid individuals by Southern blotting.

Allele 1 (12 Kb) = 0.42

Allele 2 (9 Kb) = 0.58

(Pic = 0.36)

Mendelian Inheritance: Codominant segregation demonstrated in 5 European families.

Chromosomal Localization: To localize FADJ3 relative to other DNA markers on human chromosome 6, CEPH families were studied and a two-point linkage analysis performed using the LINKAGE program (M.Lathrop) and the CEPH Public data base. FADJ3 (at the FTHP1 locus) was most closely linked to the MUT locus (lod-score: 2.87 for a recombination value of 0.01).

Probe and Sequence Availability: Contact V.David.

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Reference: 1) Dugast, I. *et al.* (1990) *Genomics* 6, 204-211.

A BamHI RFLP of the C8A gene

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Source/Description: A C8A cDNA clone was isolated from a size selected (>2 kb) human liver cDNA library and cloned into PAT153/PvuII/8 (1).

Polymorphism: BamHI identifies a four allele polymorphism (A4: 3.5 kb, A1: 3.6 kb, A3 and A2 approximately 3.53 and 3.57 kb, respectively). In addition, there are constant bands of 3.8 and 8.6 kb.

Frequency: Estimated frequency from 64 unrelated Caucasians:

A3: 0.77

A1: 0.18

A4 and A2 combined: 0.05

PIC = 0.331

Not Polymorphic For: EcoRI, HindIII, SacI, PvuII, BglII.

Chromosomal Localization: C8A has previously been mapped to 1p36.2-p22.1 (2, 3).

Mendelian Inheritance: Autosomal codominant segregation of the BamHI RFLP was observed in 33 two-generation families with 62 offspring.

Probe Availability: Contact J.S.

Other Comments: The presence of as many as four different alleles as well as the finding of successive differences of about 30 base pairs between the alleles, may indicate a VNTR polymorphism.

References: 1) Rao, A.O., Howard, O.M.Z., Ng, S., Whitehead, A.S., Colten, H. and Sodetz, J.M. (1987) *Biochem.* 26, 3556-3564. 2) Rogde, S., Olaisen, B., Gedde-Dahl, T.jr. and Teisberg, P. (1986) *Ann. Hum. Genet.* 50, 139-144. 3) Kaufman, K.M., Snider, J.V., Spurr, N.K., Schwartz, C.E. and Sodetz, J.M. (1989) *Genomics* 5, 475-480.

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