MspI RFLP in the human fumarylacetoacetate hydrolase (FAH) gene

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Source and Description of Probe: The HA2 probe is a 1.5 kb human fumarylacetoacetate hydrolase full-length cDNA coding fragment isolated from a human liver λgt11 cDNA library and subcloned into the EcoRI site of plasmid pGEM-7Zf (1).

Polymorphism: MspI resolves a two-allele polymorphism with presence (E1) or absence (E2) of a 2.4 kb fragment. Three constant bands of 3.2 kb, 1.4 kb and 1.1 kb are also seen.

Frequency: Determined from 28 unrelated French-Canadian individuals.
E1: .55
E2: .45
Heterozygosity for E1/E2 alleles is 0.50. Distribution of homozygotes and heterozygotes was consistent with Hardy-Weinberg equilibrium.

Not Polymorphic For: BamHI, HaeIII, HindIII, HpaII and PalI tested on leukocyte DNA from 15 French-Canadians including one hereditary tyrosinemia (type 1) obligate carrier and one affected child.

Also Polymorphic For: TaqI, KpnI, RsaI and BglII RFLPs described in *Nucleic Acids Res.* **19**, 1352 and 1965.

Chromosomal Localization: 15q23−25 (1).

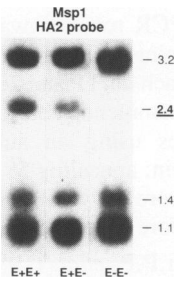
Mendelian Inheritance: Shown in fourteen families (53 individuals).

Probe Availability: Contact Dr R.M.Tanguay.

Clinical Relevance: Type 1 hereditary tyrosinemia is an autosomal recessive disease characterized by a deficiency of the enzyme fumarylacetoacetate hydrolase (2).

Acknowledgements: This work was supported by grants from the MRC of Canada and the Téléthon des Etoiles to RMT. SD holds a studentship from the MRC of Canada.

References: 1) Phaneuf,D. *et al.* (1991) *Am. J. Hum. Genet.* **48**, 525−535. 2) Tanguay,R.M. *et al.* (1990) *Am. J. Hum. Genet.* **47**, 308−316.



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Properties of a highly polymorphic locus (D2S92) located in the telomeric region of chromosome 2

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Source/Description: The probe used to detect this polymorphic locus is a 70 bp synthetic oligonucleotide corresponding to the consensus sequence found in the clone VTR1.1. The sequence of the 70 bp repeats found in this clone has been described by Krowczynska *et al.* 1990.

Polymorphisms: HaeIII identifies a VNTR-type of polymorphism. Analysis of unrelated individuals shows a continuous distribution of DNA fragments varying in size from about 2 to 13 Kb. The same type of polymorphism is also identified in DNA digested with HinfI or PstI restriction endonucleases.

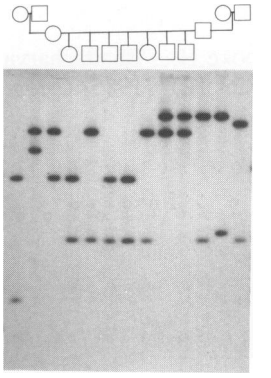
Heterozygosity: With HaeIII, 95% heterozygosity was observed in 126 Caucasians.

Chromosomal Localization: Chromosomal localization to 2p24-pter was established by hybridization to DNA from a panel of human/rodent somatic cell hybrids (mapping panel #1, NIGMS, Camden, NJ) and linkage analysis to genetic makers in chromosome 2.

Mendelian Inheritance: Co-dominant segregation demonstrated by the analysis of 32 C.E.P.H. families.

Probe Availability: DNA sequence information needed to synthesize this probe was described by Krowczynska *et al.*

Reference: 1) Krowczynska,A.M., Rudders,R.A. and Krontiris,T.G. (1990) *Nucl. Acids Res.* **18**, 1121−1127.



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