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

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).

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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 Hinfl 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.