An SstI RFLP at the C-Kit oncogene locus (KIT)

Diana Curtis
Sub-Department of Medical Genetics, University of Sheffield, 117 Manchester Road, Sheffield S10 5DN, UK

Source/Description: A 0.7 Kb SacI−SalI V-kit fragment subcloned into pUC 12 (1).

Polymorphism: The V-kit probe identifies a 2 allele SstI polymorphism with bands at > 12 Kb (Allele B1) and 6.8 Kb (Allele B2); a constant band at > 15 Kb is also present. This probe also detects a HindIII RFLP (2) which is not in linkage disequilibrium with the SstI RFLP.

Frequency: Estimated from 30 unrelated Caucasians.
Allele B1 0.84
Allele B2 0.16

Not Polymorphic For: AvaI, BanI, BstNI, EcoRV, KpnI, MboI, PvnII, KbaI with DNA from 20 pooled Caucasians in addition to the RE's reported in (2). MspI not polymorphic on this screen (2).

Chromosomal Localisation: 4p11-q22 [HGM 10].

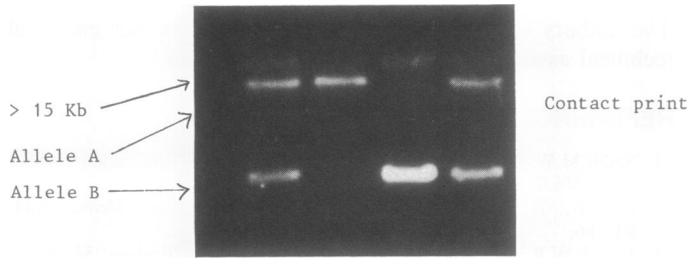
Mendelian Inheritance: Codominant inheritance in a 3 generation family of 12 members.

Probe Availability: See (1).

Other Comments: Comparison of the RFLP pattern observed in pooled human DNA with that in the DNA of a homozygous hydatiform mole is a valuable method of screening large numbers of restriction enzymes for RFLPs.

Acknowledgement: I am grateful to Dr Peter Besmer for his kind gift of the V-Kit probe.

References: 1) Besmer,P. *et al.* (1986) *Nature* **320**, 415−421. 2) Berdahl,L.D. *et al.* (1988) *Nucl. Acids Res.* **16**, 4740.



TaqI RFLP for the human fumarylacetoacetate hydrolase (FAH) gene

Sylvie I.Demers, Daniel Phaneuf and Robert M.Tanguay
Ontogénèse et Génétique Moléculaire, Centre de Recherches du CHUL, 2705 Boul. Laurier, Ste-Foy, Québec, G1V 4G2, Canada

Source/Description of Clone: The phHA2 probe is a 1.5 kb human fumarylacetoacetate hydrolase cDNA clone isolated from a human liver λgt11 cDNA library (1). This cDNA insert contains all the FAH coding sequence. The phHAB probe is a 0.583 kb SphI/PstI cDNA subfragment coding for amino acid Ala^{134} to Gln^{328}.

Polymorphism: With probe phHA2, TaqI identifies a two-allele polymorphism with fragments of 8.0 kb (A1) and 5.4 kb + 2.6 kb (A2). Four invariant bands are seen at 5.4 kb (fainter than the 5.4 kb allelic fragment), 3.4 kb, 1.9 kb and 1.3 kb. Probe phHAB reveals the same polymorphism but without the 5.4 kb constant band.

Frequency: Calculated from 35 unrelated (at least at the second degree) French-Canadian individuals.
A1 : .29
A2 : .71
Heterozygosity for A1/A2 alleles is 0.41. Observed frequencies of heterozygotes and homozygotes fit Hardy-Weinberg equilibrium.

Not Polymorphic For: BamHI, HaeIII, HindIII, HpaII and PalI studied in 15 unrelated individuals. The DNA source is blood lymphocytes.

Chromosomal Localization: The human FAH gene has been assigned to 15q23−25 (1).

Mendelian Inheritance: Codominant segregation was found in seventeen families (62 individuals).

Probe Availability: Available for collaboration. Contact Dr R.M.Tanguay.

Clinical Relevance: Fumarylacetoacetate hydrolase is the enzyme deficient in type 1 hereditary tyrosinemia (2).

Acknowledgements: Supported by the MRC of Canada (Grant PG-35) and by studentships from FRSQ and FCAR.

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