RFLP for a human cytochrome P-450 gene at 19q13.1 – qter (HGM8 provisional designation CYPI)

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Source and description of clone: A 1.2 kb cDNA inserted into the PstI site of pKT218 isolated from an adult human liver cDNA library (Phillips et al., 1985a).

Polymorphism: SstI (GAGCT/C) B.R.L. Ltd.) identifies a two allele system with a band at 9.3 kb (A1) or bands at 5.2 kb and 4.1 kb (A2). Invariant bands at 10.6 kb, 6.6 kb, 4.4 kb, 3.1 kb 2.5 kb and 0.8 kb are also identified.

Frequency: Studied in 30 European whites.
9.3 kb allele (A1) 0.55
5.2 kb and 4.1 kb allele (A2) 0.45

Not Polymorphic for: AluI, ApaI, BamHI, BclI, BglI, BglII DdeI, DpnI, EcoRI, FnDII, HhaI, HindIII, HinfI, KpnI, MboI, MspI, PstI, PvuII, RsaI, SphI, StuI, TaqI, XbaI. Screened on a panel of at least 9 individuals.

Chromosomal localisation: Probe localised to 19p13 - qter using panel of somatic cell hybrids and to 19q13.1 - qter by in situ hybridisation to fixed metaphase chromosomes. (Phillips et al., 1985b).

Mendelian inheritance: A codominant segregation pattern has been observed in 13 families.

Probe availability: Available for collaborative studies on cytochrome P-450; freely available for all other studies except for those on cystic fibrosis and Myotonic dystrophy. Correspondence should be addressed to I.R.P. or E.A.S.

Other Comments: No evidence of repeat sequence hybridisation. Low background at a stringency of 1xSSC.

References: Phillips et. al. (1985a) - Proc. Natl. Acad. Sci. 82:983-987.
Phillips et. al. (1985b) - Annals of Human Genetics in press.

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