A TaqI RFLP identified at the retinoblastoma locus on chromosome 13

R. Shiang, J.C. Murray, J. Wiggs¹ and T. Dryja¹

Department of Pediatrics, University of Iowa, Iowa City, IA 52242 and ¹Massachusetts Eye and Ear Infirmary, Boston, MA 02114, USA

SOURCE/DESCRIPTION: Probe D95HS0.5 is a 0.6 kb fragment subcloned into Bluescribe a pUC19 derivative from a bacteriophage library isolated by a cDNA probe of the retinoblastoma gene¹. The fragment is released with the enzymes HindIII and SalI.

POLYMORPHISMS: TaqI identifies a 2 allele polymorphism with a band at 2.1 kb and 1.8 kb with a frequency of 0.97 and 0.03 respectively. There are no constant bands.

NOT POLYMORPHIC FOR: ApaI, BamHI, BclI, BglI, BglII, BspI286, BsmI, BstXI, DdeI, DraI, EcoI09, EcoRI, EcoRV, HaeIII, HindIII, HincII, HinfI, HphI, MboI, MboII, MspI, NaeI, NciI, NdeI, NheI, NruI, NsiI, PaeR7I, PflMI, PstI, PvuII, RsaI, SacI, SacII, Sau3AI, Sau96I, ScaI, SpeI, SspI,* (see below)

CHROMOSOMAL LOCALISATION: Probe assigned to chromosome 13 using a linkage analysis with retinoblastoma¹.

MENDELIAN INHERITANCE: Codominant inheritance shown in 3 CEPH pedigrees

PROBE AVAILABILITY: Contact T. Dryja

OTHER COMMENTS: A KpnI RFLP was previously reported with allele sizes of 12.0 and 8.0 and a frequency of 0.95 and 0.05 respectively. *Also not polymorphic for StuI, XbaI, XhoI, XmnI tested in 7 to 10 unrelated Caucasians.

REFERENCE: 1 J. Wiggs et al. 1988. New Engl J Med 318:151-157