

PCR detection of the BglII RFLP at the RBP3 locus

Joseph A. Rodriguez, Greg I. Liou¹ and Stephen P. Daiger
Graduate School of Biomedical Sciences, The University
of Texas Health Science Center, Houston, TX and ¹Cullen
Eye Institute, Baylor College of Medicine, Houston, TX,
USA

Source/Description: A 2184 bp cDNA fragment designated H-4IRBP was previously reported to detect a polymorphism at the RBP3 locus (1). We have amplified the first intervening sequence (IVS1) of the RBP3 gene using exon to exon amplification and PCR (2). We have used this product as a hybridization probe to enhance detection of the polymorphism (see Fig. 1). Using this probe we have been able to map the polymorphism to IVS2 (see fig. 2).

Polymorphism: BglII (AGATCT) identifies a two allele polymorphism with bands at 6.3 kb and 4.3 kb. No invariant bands are detected by this method.

Chromosomal Location: 10q11.2.

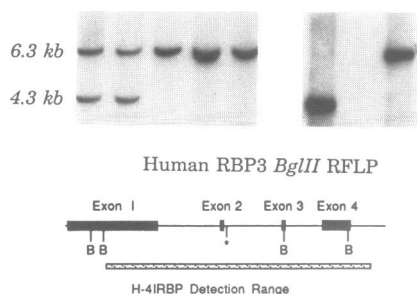
Mendelian Inheritance: Demonstrated in a large multi-generation family (3).

Probe Availability: Not needed for PCR amplification. Contact G. Liou for H-4IRBP probe.

Other Comments: Oligonucleotides used for amplification are 5'-CATTCTGGGAATTGTGCCCA-3' and 5'-CAAATACTT-CAGGGGAAGGG-3'. These sequences are situated at the 3' end of exon 1 and the 5' end of exon 2 respectively. The amplification is done using Taq polymerase under the conditions recommended by the manufacturer (Perkin Elmer-Cetus). For 25 cycles at 94°C 1 min, 57°C 50 sec and 72°C for 1.5 min. An aliquot of the amplification product is electrophoresed on a low-melt agarose gel. The resulting 1.8 kb band is excised from the gel, labeled using the hexamer labeling method, and used as a hybridization probe (4).

Acknowledgements: Supported by grants from the National Retinitis Pigmentosa Foundation and George Gund Foundation (S.P.D.), the National Retinitis Pigmentosa Foundation (G.I.L.), NIH Grant EY07142 (S.P.D.) and NIH Grant EY03829 (G.I.L.).

References: 1) Liou, G.I. *et al.* (1987) *Nucl. Acids Res.* **15**, 3196. 2) Liou, G.I. *et al.* (1989) *J. Biol. Chem.* **264**, 8200–8206. 3) Daiger, S.P. *et al.* (1989) *Cytogenet. Cell Genet.* **50**, 181–187. 4) Feinberg, A.P. *et al.* (1983) *Anal. Biochem.* **132**, 6.



BanI polymorphism at the XBP1 locus

Bertrand Fontaine, Mark P. Hanson, Hsiou-Chi Liou¹,
Laurie H. Glimcher¹, Guy A. Rouleau² and
James F. Gusella

Molecular Neurogenetics Laboratory, Neuroscience
Center, Massachusetts General Hospital and Harvard
Medical School, Boston, MA 02114, ¹Department of
Cancer Biology, Harvard School of Public Health, Boston,
MA 02115, USA and ²Centre de Recherche en
Neurosciences, McGill University, The Montreal Hospital
Research Institute, Montreal, Quebec H3G 144, Canada

Source/Description: A 0.9 kb EcoRI-fragment of the human X-box binding protein cDNA subcloned in pUC18 (1).

Polymorphism: BanI identifies a two allele-polymorphism (allele 1: 8.5 kb, allele 2: 7.5 kb).

Frequency: Estimated from 16 chromosomes of unrelated individuals:

1: 0.70

2: 0.30

Not Polymorphic For: XmnI, BstXI, TaqI, HincII, PstI, KpnI, EcoRV, BclI, HindIII, BglI, BanII, BamHI, ApaI, SacI, MspI, BglII, HinfI, Sau96I, ScaI, StuI, RsaI, SauIIIa, PvuII, BclI.

Chromosome Location: The BanI polymorphism has been assigned to chromosome 5 using somatic cell hybrids.

Mendelian Inheritance: Co-dominant segregation of the BanI polymorphism was observed in 10 two generation families of the Venezuela Reference Pedigrees (2).

Probe Availability: Contact Dr L.H. Glimcher.

References: 1) Liou, H.C. *et al.* (1990) *Science* **247**, 1581–1584. 2) Tanzi, R.E. *et al.* (19??) *Genomics* **3**, 129–136.

