

An EcoO109 RFLP for the SNRPE gene on chromosome 1

B.L.Miller, D.Y.Nishimura, E.D.Wieben[1] and J.C.Murray*
Department of Pediatrics, University of Iowa, Iowa City, IA 52242 and [1]Department of Biochemistry and Molecular Biology, Mayo Clinic, Rochester, MN 55905, USA

Source/Description: A 1400 bp BamHI/BglII human genomic fragment containing 1891−3296 of the small nuclear ribonucleoprotein polypeptide E (SNRNPE) gene cloned into Bluescribe (p882EBBg-4) (1). This fragment contains all of exon 2 and portions of introns 1 and 2. No known repetitive elements are present in this clone.

Polymorphism: EcoO109 detects polymorphic fragments of 4.6 kb (B1) and 2.3 kb (B2).

Frequency: The allele frequencies were determined by typing 80 unrelated individuals of the CEPH reference panel.

Enzyme	Allele	Fragment Size	Frequency
EcoO109	B1	4.6 kb	0.18
	B2	2.3 kb	0.82

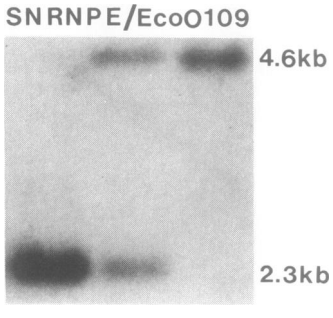
Not Polymorphic For: AluI, AlwNI, BamHI, BanII, BglI, BglII, Bsp1286, BspHI, DdeI, EcoRI, EcoRV, HaeIII, HincII, HindIII, HinfI, KpnI, NheI, NsiI, PstI, PvuII, RsaI, SacI, SspI, StuI, XbaI, XmnI.

Chromosomal Localisation: Mapped to 1q25-q43 using somatic cell hybrids (2) and linked to Renin (Nishimura, unpublished results).

Mendelian Inheritance: Co-dominant segregation observed in 22 CEPH families.

Probe Availability: Available from E.D.Wieben.

References: 1) Stanford,D.R., Perry,C.A., Holicky,E.L., Rohleder,A.M. and Wieben,E.D. (1988) *J. Biol. Chem.* **263**, 17772−17779. 2) Neiswanger,K., Stanford,D.R., Sparkes,R.S., Nishimura,D., Mohandas,T., Klisak,I., Heinzman,C. and Wieben,E.D. (1990) *Genomics* **1**, 503−508.



* To whom correspondence should be addressed

D12S17 and D12S25 identify the same MspI RFLP

Anne E.Hughes
Department of Medical Genetics, The Queen's University of Belfast, Belfast City Hospital, Belfast BT9 7AB, UK

Source/Description: Probe pYNH15 (D12S17) is a 3.2 kb fragment of human genomic DNA cloned into pUC18 (1). Probe CRI-L809 (D12S25) consists of a fragment of human genomic DNA of unknown size cloned into lambda Charon 4A (2).

Polymorphism: Both probes detect an identical MspI three allele system (A1 = 4.0 kb, A2 = 3.2 kb, A3 = 2.65 kb).

Frequency: Estimated from 14 unrelated Caucasians.
A1 = 0.11
A2 = 0.75
A3 = 0.14

Chromosomal Localization: HGM10 localized D12S17 to chromosome 12q and D12S25 to chromosome 12.

Mendelian Inheritance: Identical segregation of codominant alleles was observed in one large kindred (51 individuals).

Probe Availability: pYNH15 is available from ATCC, and CRI-L809 from Collaborative Research Inc.

Other Comments: Triplicate blots of MspI-digested DNA from six unrelated individuals were hybridized with either pYNH15 or CRI-L809 or a mixture of both probes. The same polymorphism was detected by both probes (Fig. 1). CRI-L809 also identified constant bands at 1.65 kb, 1.3 kb and 1.15 kb. No additional bands were shown when both probes were hybridized simultaneously, thus confirming that D12S17 and D12S25 represent the same locus.

Acknowledgement: This study was supported by a grant from the Wellcome Trust.

References: 1) O'Connell,P. *et al.* (1987) *Genomics* **1**, 93−102. 2) Donis-Keller,H. *et al.* (1987) *Cell* **51**, 319−337.