
MspI RFLP for SNRNPE gene on 1q

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SOURCE/DESCRIPTION: A 1400bp 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: MspI detects polymorphic fragments of 1.4kb (A1) and 1.1kb (A2).

FREQUENCY:

A1 = .10

A2 = .90 in 122 unrelated Caucasian individuals of the CEPH reference panel

NOT POLYMORPHIC FOR: AluI, AlwNI, BamHI, BanII, BglI, BglIII, 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, unpub. results)

MENDELIAN INHERITANCE: in 40 families of the CEPH reference panel

PROBE AVAILABILITY: Contact E.D. Wieben

REFERENCE:

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., Wieben E.D., Mohandas T., Klisak I., Heinzmann C., and Sparkes R.S. (1988) Am J Hum Genet 43:A153 abst. 8.104 suppl.

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