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A PvuII RFLP detected in the human prion protein (PrP) gene

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SOURCE AND DESCRIPTION OF CLONE: Probe pEA974 (1), contains a 974 bp fragment present in PrP clone XIV inserted into pBR322. The probe was sublocalized to chromosome 20p (2).

POLYMORPHISM: PvuII identifies a two allele polymorphism of either a band at 1.5 Kb or two bands at 1 and 0.5 Kb.

FREQUENCY: Studied in 120 European Caucasians:

1.5 Kb allele (A1):	0.10
1 & 0.5 Kb allele (A2):	0.90

NOT POLYMORPHIC FOR: (studied in 20 unrelated European Caucasians) - ApaI, ApaLI, AvaI, AvuI, BanI, BamHI, BclI, BglI, BglII, BstEII, BstNI, EcoRI, EcoRV, HaeIII, HindIII, HinfI, KpnI, MspI, NheI, NruI, PstI, RsaI, SmaI, SstI, StuI, TaqI, XbaI, and XmnI.

MENDELIAN INHERITANCE: Autosomal codominant inheritance was shown in two informative families. One is illustrated below.

PROBE AVAILABILITY: Contact Nikolaos K. Robakis.

OTHER COMMENTS: There is a linkage between an XbaI RFLP for PrP in the mouse and scrapie incubation time genes (3). We predict a similar linkage in familial human dementias, such as C-J disease and GSS, which may relate to slow virus susceptibility.

REFERENCES: (1) Robakis et al (1986) PNAS 83: 6377-6381.
(2) Robakis et al (1986) BBRC 140:758-765.
(3) Carlson et al (1986) Cell 46: 503-511.

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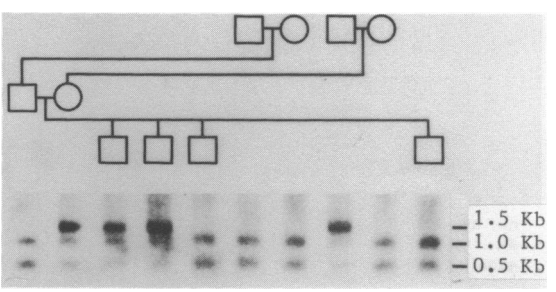

Figure 1. A family showing autosomal codominant inheritance of the PvuII RFLP. The mother was heterozygous and two of four sons received the 1.5 Kb allele (A1).

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