A high frequency NcoI RFLP detected in the Alzheimer amyloid peptide gene

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SOURCE/DESCRIPTION: Probe pAMB2.3 contains a 778bp cDNA fragment encoding the amyloid polypeptide of Alzheimer disease and Down Syndrome (beta protein) inserted into pUC18 (1). The probe was sublocalized to chromosome 21q21 (2).

POLYMORPHISM: NcoI identifies a two allele polymorphism of a band at either 15kb or 8.5 kb.

FREQUENCY: Studied in 41 unrelated individuals:

15kb allele (A1): 0.67

8.5kb allele (A2): 0.33

NOT POLYMORPHIC FOR: AluI, AvaI, ApaI, ApaLI, BamHI, BanI, BclI, BglI, BglII, BstEII, ClaI, EcoRV, HaeIII, HhaI, HinfI, HinPI, KpnI, MboI, MspI, NaeI, NarI, NciI, NcoI, NheI, PaeR71, PstI, RsaI, SacII, SstI, StuI, StyI, TaqI, XbaI, XhoI, XmnI

MENDELIAN INHERITANCE: Autosomal codominant inheritance was shown in five informative families.

PROBE AVAILABILITY: Contact Nikolaos K. Robakis

OTHER COMMENTS: Two non polymorphic bands, 7.0kb and 6.0kb, are seen only occasionally. Their presence seems to depend on the particular DNA preparation.

REFERENCES: (1) Robakis et al. (1987) PNAS 84: 4190-4194
(2) Robakis et al. (1987) Lancet 1: 384-385

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