Isolation and mapping of a polymorphic DNA sequence pEFD4.2 on chromosome 19 [D19S22]

Y.Nakamura, E.Fujimoto, L.Ballard, P.O'Connell, M.Leppert, G.M.Lathrop, J.-M.Lalouel and R.White

The Howard Hughes Medical Institute, University of Utah Medical School, Salt Lake City, UT 84132, USA

SOURCE/DESCRIPTION: A 3.5 kb PstI fragment from cosmid EFD4 isolated by HBV-4 oligonucleotide (1) was subcloned into the PstI site of pUC18.

POLYMORPHISM: PvuII identifies a 2 allele VNTR polymorphism with a band at 2.4 kb (P1) or 2.2 kb (P2) without a constant band. MspI, TaqI, and PstI also identify the same polymorphism.

FREQUENCY: Estimated from 94 unrelated Caucasians.

PvuII P1 : 0.38

P2 : 0.62

NOT POLYMORPHIC FOR: BglII

CHROMOSOMAL LOCALIZATION: pEFD4.2 has been assigned to chromosome 19 by multipoint linkage analysis(2) with loci (LDLR, APOC2) known to span this region(3,4).

MENDELIAN INHERITANCE: Co-dominant segregation of the PvuII RFLP was observed in 47 three generation families.

PROBE AVAILABILITY: Contact Y.N.

OTHER COMMENTS: RFLPs were observed under normal hybridization and wash stringencies.

REFERENCES:

1. Y. Nakamura et al., Science 235: 1616-1622 (1987)
2. G.M. Lathrop et al., Am. J. Hum. Genet. 37:482-498 (1985)
3. S. Naylor et al., Cytogenet. Cell Genet. 40:242-267 (1985)
4. Y. Nakamura et al., abstract submitted to Human Gene Mapping Workshop 9. Cytogenet. Cell Genet., in press