A hypervariable locus D16S309 located at the distal end of 16p

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Source/Description: Probe MS205 was detected by probe 33.15 (1) in a lambda L47.1 library of *Mbo*I digested, size fractionated DNA (5–20kbp) pooled from 20 unrelated individuals (2). The 5.1kbp insert was subcloned into the *Bam*HI site of pUC13 resulting in the subclone pMS205.2 which was propagated in *E.coli* strain DH5 α (BRL).

Polymorphism: pMS205.2 detects a hypervariable locus in genomic DNA digested with *Mbo*I and *Hinf*I revealing DNA fragments 4–7kbp and 2–5kbp in length respectively. It cannot be detected in *Hae*III, *Rsa*I or *Hpa*II genomic digests as these enzymes cut within the minisatellite repeat unit.

Heterozygosity: The D16S309 locus has multiple alleles and a heterozygosity of 96% or 97% among 202 unrelated Caucasians and 204 unrelated Afro-Caribbeans tested respectively.

Chromosome Localization: The D16S309 locus has been assigned to chromosome 16 by hybridization of the probe pMS205.2 to a human-rodent somatic cell hybrid panel of DNAs and to 16p by linkage analysis, where it is closely linked to the 3' and 5' HVRs of the α -globin locus which is located approximately 145kbp from the telomere of 16p (3).

Mendelian Inheritance: Co-dominant inheritance of the *Mbo*I DNA fragments detected by pMS205.2 has been shown among the 40 reference families of the CEPH panel.

Other Comments: The D16S309 locus has a germline mutation rate to new length alleles of 0.004 per gamete, estimated from 257 families where maternity and paternity have been verified.

Probe Availability: pMS205.2 is available for research purposes and can be obtained from Cellmark Diagnostics (8 Blacklands Way, Abingdon Business Park, Abingdon, OX14 1DY, UK).

Acknowledgements: We thank Dr S.Povey for generously donating the human-rodent somatic cell hybrid panel of DNAs and CEPH for providing the panel of 40 family DNAs. NJR is an HGMP Senior Fellow; JALA is a Wellcome Trust Postdoctoral Fellow. This work was funded by grants from the MRC and HGMP.

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A dinucleotide repeat polymorphism in the human Na⁺,K⁺ ATPase, alpha subunit (ATP1A3) gene

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Source/Description: The polymorphic dinucleotide repeat was isolated and characterized from cosmid 67H9. Cosmid 67H9 was obtained by screening the LNL chromosome 19 cosmid library with the human Na⁺,K⁺ ATPase cDNA (ATP1A3) (1). Sau 3A1 fragments were subcloned into pUC19 and selected by hybridization to poly (dC-dA). Predicted length of the amplified sequence was 172 bp.

Primer Sequences:

Primer 1: ATG CCT GCT CCC CTA CAT GA

Primer 2: GAT GGA CAC CAG ATG ACC TC

Frequency: Estimated from 100 chromosomes from 50 unrelated individuals. PIC = 0.54 and HET = 0.57.

Allele	(bp)	Frequency
C1	176	0.01
C2	174	0.01
C3	172	0.11
C4	170	0.11
C5	168	0.62
C6	166	0.10
C7	164	0.03

Chromosomal Localization: The human Na⁺,K⁺ ATPase, alpha subunit (ATP1A3) has been assigned to chromosome 19q12-q13.2 (1). The primers were also tested against a human chromosome localization panel (BIOS) and assigned to chromosome 19.

Mendelian Inheritance: Co-dominant inheritance was observed in three CEPH families.

Protocol: PCR was performed with 30 ng genomic DNA and 60 ng of each primer (2). The samples were processed through 27 cycles of 94° for one minute, 55° for 2 minutes, and 72° for one minute. The products were run on a 6.5% denaturing polyacrylamide gel.

Acknowledgements: We are grateful to Drs.H.G.Harley and D.E.Housman for providing the cDNA clone. This work was supported by NIH grant NS 19999 (ADR) and a Leadership and Excellence in Alzheimer's Disease (LEAD) Award AGO 7922 (ADR).

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