90-5

A polymorphic DNA probe from chromosome 7 (7q22)

T.Casals, V.Nunes and X.Estivill[1]
Molecular Genetics Department, Fundació Investigació Santa Creu i Sant Pau, 08025 Barcelona, Spain and [1]Molecular Genetics Department, St Mary's Hospital Medical School, London W2 1PG, UK

Source/Description: p6a-1 is a 0.9 kb HindIII/BamHI genomic fragment subclone of cosmid cNX.6a in pUC13. cNX.6a was isolated from a non-methylated enriched library from the CMGT cell line CII (1, 2).

Polymorphism: MspI (CCGG) identifies a three allele polymorphism with bands at 2.4 kb (A1), 1.8 kb (A2) and 1.35 kb (A3).

Frequency: Studied in 88 European Caucasians:
A1: 0.02
A2: 0.41
A3: 0.57

Not Polymorphic For: BamHI, HindIII, PstI, PvuII, XbaI, EcoRI, TaqI, HincII.

Chromosomal Localisation: To chromosome 7q22 using a panel of somatic human/rodent cell hybrids (3).

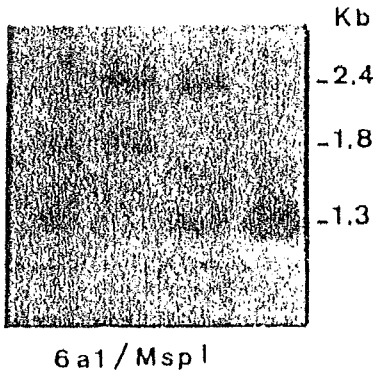
Mendelian Inheritance: Co-dominant segregation was demonstrated in 24 families.

Probe Availability: Contact X.Estivill.

Other Comments: The original cosmid clone recognizes a non-methylated CpG island.

Acknowledgements: Supported by the 'Fondo de Investigaciones Sanitarias de la Seguridad Social' (89/0563) and the Spanish Ministry of Education and Science (PB87–0074).

References: 1) Estivill *et al.* (1989) *Am. J. Hum. Genet.* **44**, 704–710. 2) Scambler *et al.* (1986) *Nucl. Acids Res.* **14**, 7159–7173.



Probe 218EP6 (D16S246) detects RFLP's close to the locus affecting adult polycystic kidney disease (PKD1) on chromosome 16

F.G.M.Snijdewint, J.J.Saris, J.G.Dauwerse, M.H.Breuning and G.J.B.van Ommen
Institute of Human Genetics, State University of Leiden, Wassenaarseweg 72, 2333 AL Leiden, The Netherlands

Source/Description: Probe 218EP6 (D16S246) contains two PstI fragments subcloned in pKUNI after PstI digestion of a cosmid (218) from a random MboI library. The 1.7 kb insert is a fragment detecting the RFLP.

Polymorphism: The probe detects a two-allele insertion/deletion polymorphism of 0.3 kb with a number of restriction enzymes.

	Allele 1 (Ha)	Allele 2 (Hb)
EcoRI	7.1 kb	6.8 kb
BamHI	3.7 kb	3.3 kb
PvuII	2.8 kb	2.5 kb
RsaI	2.6 kb	2.3 kb
PstI	1.5 kb	1.2 kb
TaqI	1.2 kb	0.9 kb

The probe also detects a two-allele polymorphism for BglII with fragment sizes 9.6 kb (allele 1) and 6.4 kb (allele 2) and for MspI with fragment sizes 2.0 kb (allele 1) and 1.3 kb (allele 2).

Minor variations were detected in the fragment sizes indicating that the probe may be located close to a VNTR. These variations, especially in Hb, make the probe even more informative in a number of families.

Frequency: The allele frequencies were studied in 41 unrelated individuals. Fragment 1 (Ha) 0.36 and fragment 2 (Hb) 0.64.

Not Polymorphic For: HindIII and SacI.

Chromosomal Localization: The probe is localized proximal to CMM65 (D16S84) and distal to breakpoint GM2324 t(16;22)(p13.3;q21) (3).

Mendelian Inheritance: The polymorphism segregates in a Mendelian fashion in a large number of families with PKD1.

Other Comments: The probe is scrambled with mouse DNA.

Probe Availability: The probe is available for diagnostic purposes.

Acknowledgement: This work was supported by a grant from the Netherlands Kidney Foundation.

References: 1) R.N.H.Konings *et al.* (1986) *Gene* **46**, 269–276. 2) D.F.Callen (1986) *Ann. Genet.* **29**, 235–239. 3) M.H.Breuning *et al.* (1989) *Cytogenet. Cell Genet.* **51**, 969.