CA repeat polymorphism at the D5S82 locus, proximal to adenomatous polyposis coli (APC)

C.Breukel, C.Tops, C.van Leeuwen, H.van der Klift, Y.Nakamura[1], R.Fodde* and P.M.Khan
Department of Human Genetics, Leiden University, The Netherlands and [1]Cancer Institute, Tokyo 170, Japan

Source and Description: The polymorphic CA repeat CAYN5.64c was isolated and characterized from cosmid cYN5.64 (D5S82) (1). From the same cosmid a PstI and a TaqI RFLP were previously characterized (1).

Primer Sequence:
CA primer = YN5.64c.CA 5'-CCCAATTGTATAGATTTA-GAAGTC-3'
TG primer = YN5.64c.TG 5'-ATCAGAGTATCA-GAATTTCT-3'

Polymorphism and Frequency: Six alleles were detected in a sample of 51 unrelated Dutch volunteers. PIC = 0.70

Allele	Number of Ca repeats	Frequency	Product size (bp)
C1	15	0.02	179
C2	14	0.26	177
C3	13	0.30	175
C4	12	0.02	173
C5	11	0.16	171
C6	10	0.24	169

Mendelian Inheritance: Autosomal co-dominant segregation was observed in 25 Dutch families.

Clinical Relevance: Presymptomatic diagnosis of familial adenomatous polyposis (FAP).

Chromosomal Localization: Assigned to the long arm of chromosome 5(q15-q23) between markers D5S71 and D5S81 by linkage analysis (1) and by fluorescent in situ hybridization (2).

Other Comments: The products of the PCR reactions are loaded on 10% denaturing (8M urea) polyacrylamide gels in TAE buffer. The electrophoresis is performed overnight at 12.5 V/cm in an 'aquarium tank' filled with buffer and maintained at 60°C. The gel is then stained with ethidium bromide and the bands are directly observed under U.V. light.

References: 1) Nakamura,Y. *et al.* (1989) *Am. J. Hum. Genet.* **43**, 638–644. 2) Tops,C.M.J. *et al.*, in preparation.

* To whom correspondence should be addressed

*Nco*I RFLP at 19q13 identified by the DNA sequence pW119B (D19S169)

C.Lavedan, D.Savoy, M.Simon, B.Wieringa[1] and C.Junien*
INSERM U 73, Château de Longchamp, Bois de Boulogne, 75016 Paris, France and [1]Department of Cell Biology, University of Nijmegen, 6500 HB Nijmegen, The Netherlands

Source/Description: pW119B is a 0.98 kb DNA segment, subcloned in a pBluescript II SK + vector (Stratagene) and derived from a EMBL3 library, constructed from the somatic cell hybrid 20XP3542-1-4 (1). *SalI*/*SacI* double digestion releases the 0.98 kb insert.

Polymorphism: The probe pW119B detects a two-allele polymorphism with *Nco*I.

Frequency: Allele frequencies were determined by typing 22 unrelated Caucasians.

Allele	Size	Frequency
A1	8.6 kb	0.64
A2	7.8 kb	0.36

Chromosomal Localisation: Localised at 19q13.2–13.3 in a region defined proximally by the breakpoint in the hybrid 20XP3542-1-4 and distally by the breakpoint in the CKMM gene in the t (X; 19) somatic cell hybrid 908KIB18 (2).

Mendelian Inheritance: Co-dominant inheritance demonstrated in 8 Myotonic Dystrophy families.

Probe Availability: Available for collaborative work on Myotonic Dystrophy, contact C. Junien at the above address.

Acknowledgement: This work was supported by the 'Association Française contre les Myopathies'.

References: 1) Stallings *et al.* (1988) *Am. J. Hum. Genet.* **43**, 144–151. 2) Smeets *et al.* (1990) *Am. J. Hum. Genet.* **46**, 492–501.

* To whom correspondence should be addressed