

CA repeat polymorphism within the MCC (mutated in colorectal cancer) gene

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Source and Description: The polymorphic CA repeat CAMBC was isolated and characterized from cosmid cLDMBC. Cosmid cLDMBC was derived by screening the flow sorted chromosome 5 cosmid library LA05NCO1 with the MCC cDNA probe SW15 (1).

Primer Sequences:

TG primer = MBC.TG 5'-GTGTTGGCAATGCAGTAGTG-3'
CA primer = MBC.CA 5'-ACCACTGGACTCCAGCCTGG-3'

Polymorphism and Frequency: Four alleles were detected in a sample of 46 unrelated Dutch volunteers. PIC = 0.49.

Allele	Number of CA repeats	Frequency (bp)	Product size
A1	28	0.01	176
A2	26	0.18	174
A3	22	0.19	170
A4	20	0.62	168

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) by using a panel of human-Chinese hamster radiation hybrids 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.

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References: 1) Kinzler, K.W. *et al.* (1991) *Science* **251**, 1366–1370. 2) Tops, C.M.J. *et al.*, in preparation.

CA repeat polymorphism at the D5S299 locus linked to adenomatous polyposis coli (APC)

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Source and Description: The polymorphic CA repeat CACB26 was isolated and characterized from cosmid cCB26 (D5S299). Cosmid cCB26 was derived from a reduced complexity radiation hybrid which contained fragments of human chromosomes 4 and 5 in a Chinese hamster background (1).

Primer Sequence:

TG primer = CB26.TG 5'-GTAAGCAGGACAA-GATGACAG-3'
CA primer = CB26.CA 5'-GCTATTCTCTCAGGATCTTG-3'

Polymorphism and Frequency: Seven alleles were detected in a sample of 26 unrelated Dutch volunteers. PIC = 0.66.

Allele	Number of CA repeats	Frequency	Product size (bp)
A1	24	0.02	182
A2	23	0.02	180
A3	22	0.33	178
A4	21	0.04	176
A5	20	0.15	174
A6	19	0.04	172
A7	11	0.40	156

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) by using a panel of human-Chinese hamster radiation hybrids and by fluorescent in situ hybridization (1).

Other Comments: The products of the PCR reactions are loaded on 11% denaturing (8 M urea) polyacrylamide gels in TAE buffer. The electrophoresis is performed overnight at 12.5 V/cm in an 'aquarium tank' filled with buffer maintained at 60°C. The gel is then stained with ethidium bromide and the bands are directly observed under U.V. light.

Reference: 1) Tops, C.M.J. *et al.*, in preparation.

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