Atlas of Genetics and Cytogenetics
in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Gene Section
Short Communication

APC (adenomatous polyposis coli)

Richard Hamelin

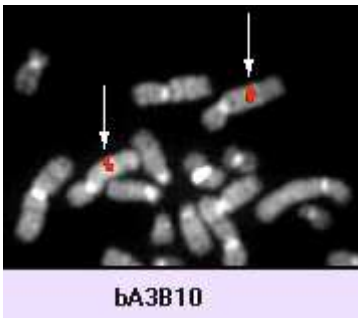
INSERM U434, Laboratoire de Genetique des Tumeurs, CEPH, 27 rue Juliette Dodu, 75010 Paris, Paris

Published in Atlas Database: April 1998

Online updated version: http://AtlasGeneticsOncology.org/Genes/APC118.html
DOI: 10.4267/2042/37429

Identity

Location: 5q21



APC (5q21) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics. Laboratories willing to validate the probes are welcome: contact rocchi@biologia.uniba.it.

DNA/RNA

Description

15 exons (with a particularly large 15th exon).

Transcription

9.0 kb mRNA; 8538 bp open reading frame.

Protein

Description

2843 amino acids; 300 kDa.

Function

Tumour suppressor gene; the APC normal gene product interacts with the adherens junction proteins a and beta-catenin suggesting it is involved in cell adhesion; since beta catenin activate transcription by forming complexes with members of the Tcf-Lef family, it is supposed that APC, by complexing beta-catenin, may also inhibit the transcription pathway regulated by the beta-catenin/Tcf complex.

Homology

Limited functional homology to known proteins.

Mutations

Germinal

In familial adenomatous polyposis.

Somatic

The APC gene is mutated in about 50% of sporadic colorectal tumours; the great majority of APC gene mutations are frameshifts of nonsense mutations resulting in premature stop codons.

Implicated in

Colorectal cancer

Disease

Adenocarcinoma; the third most frequent cancer in the world.

Prognosis

5 yr survival rate is around 50%.

Cytogenetics

There are two types, according to the ploidy: 1- aneuploid tumours showing numerous allelic losses, in particular loss of heterozygosity on chromosomes 17 and 18, and 2- diploid tumours without frequent allelic losses.

Oncogenesis

A number of genes (oncogenes and tumour suppressor genes) are known to be implicated in tumour progression in colorectal cancers; other genes are: p53, Ki-ras, and mismatch repair genes (MMR genes).

Familial adenomatous polyposis

Disease

Autosomal dominant cancer prone disease.

Prognosis

Colorectal cancer is the first cause of death in this disease.

References

Groden J, Thliveris A, Samowitz W, Carlson M, Gelbert L, Albertsen H, Joslyn G, Stevens J, Spirio L, Robertson M, Sargeant L, Krapcho K, Wolff E, Burt R, Hughes JP, Warrington J, McPherson J, Wasmuth J, Le Paslier D, Abderrahim H, Cohen D, Leppert M, White R. Identification and characterization of the familial adenomatous polyposis coli gene. Cell 1991;66:589-600.

Nishisho I, Nakamura Y, Miyoshi Y, Miki Y, Ando H, Horii A, Koyama K, Utsunomiya J, Baba S, Hedge P, Markham A, Krush AJ, Petersen G, Hamilton SR, Nilbert MC, Levy DB, Bryan TM, Preisinger AC, Smith KJ, Su LK, Kinzler KW, Vogelstein B. Mutations of chromosome 5q21 genes in FAP and colorectal cancer patients. Science 1991;253:665-669.

Powell SM, Zilz N, Beazer-Barclay Y, Bryan TM, Hamilton SR, Thibodeau SN, Vogelstein B, Kinzler KW. APC mutations occur early during colorectal tumorigenesis. Nature 1992;359:235-237.

Kinzler KW, Vogelstein B. Lessons from hereditary colorectal cancer. Cell 1996;87:159-170.

This article should be referenced as such:

Hamelin P. APC (adenomatous polyposis coli). Atlas Genet Cytogenet Oncol Haematol.1998;2(3):75-76.