APC (adenomatous polyposis coli)

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Identity

Location: 5q21

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

This article should be referenced as such: