| PublisherInfo | | |
|---|---|---|
| PublisherName | : | BioMed Central |
| PublisherLocation | : | London |
| PublisherImprintName | : | BioMed Central |

# *APC* gene mutations in human breast cancers

| ArticleInfo | | |
|---|---|---|
| ArticleID | : | 3747 |
| ArticleDOI | : | 10.1186/bcr-2000-66710 |
| ArticleCitationID | : | 66710 |
| ArticleSequenceNumber | : | 19 |
| ArticleCategory | : | Paper Report |
| ArticleFirstPage | : | 1 |
| ArticleLastPage | : | 3 |
| ArticleHistory | : | RegistrationDate : 2000–7–10<br>OnlineDate : 2000–7–10 |
| ArticleCopyright | : | Current Science Ltd2000 |
| ArticleGrants | : | |
| ArticleContext | : | 1305833 |

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## Keywords

*APC* gene, breast, mutation, truncation

# Introduction

The importance of *APC* gene mutations in the initiation step of both familial and sporadic colorectal carcinogenesis is well known. Somatic mutations of this gene have also been implicated in pancreatic, gastric and oesophageal cancers. In contrast, only a few *APC* mutations have been reported in primary human breast cancers. The vast majority of reported *APC* mutations are protein-truncating, and a novel yeast-based screening method for detection of truncating mutations is presented here.

# Aims

To utilise a novel assay to analyse the *APC*mutations in clinical samples of colorectal, breast and lung cancers.

# Comments

Investigations into the role of the adenomatous polyposis coli (*APC*) gene in carcinogenesis have frequently found few mutations associated with breast cancer. The evidence presented here implicates the gene in a higher proportion of primary breast carcinomas than previously reported, with the intriguing observation that breast tumours show a different mutation pattern to colon cancers. Further work will be required to validate the novel methodology, and to investigate the significance of the mutations in breast tumorigenesis.

# Methods

Seventy primary breast cancer specimens were analysed by the *APC* Yeast Color Assay. This involves PCR and co-transformation of *APC* mRNA and genomic DNA into yeast with a linearised expression vector carrying the 5' and 3' ends of the *APC* cDNA. After homologous recombination and fusion protein expression, yeast transformants that contained gap-repaired plasmid were selected on a leucine deficient medium. Yeast containing cDNA (or DNA) fragment of wild-type *APC* and of mutant (truncating) *APC*formed white and red colonies, respectively.

# Results

Clonal *APC* gene mutations were confirmed in 13 out of 70 (18%) of breast cancers studied. Fourteendifferent mutations were identified, widely distributed between codons 1058 and 1795 of the *APC* coding region, and with 8 of the 14 not found in the *APC* mutation database. *APC* mutations were frequently found at the high-grade and advanced stage of primary breast cancers.

# Discussion

In the present study, mutations in the *APC*gene were detected by a novel method in 18% of primary breast cancers. This is far higher than has previously been suggested, although indirect evidence by loss of heterozygosity or lost/reduced Adenomatous polyposis coli protein expression has been reported. The authors conclude that this discrepancy may be associated with the poor sensitivity of mutation detection of previous methods.

## References

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