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Evaluation of dHPLC in mutation screening of the APC gene in a Greek FAP cohort.

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Source

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Abstract

BACKGROUND:

Germline mutations in the APC gene predispose to colorectal adenomas leading to cancer in over 80% of patients. A multitude of mutations, dispersed throughout the gene, have been described. We wanted to evaluate the usefulness of denaturing high performance liquid chromatography (dHPLC) for mutation screening.

MATERIALS AND METHODS:

Ten amplicons containing 14 mutations in the APC, previously identified by sequencing in 22 FAP patients, were analysed by dHPLC. dHPLC was also used to screen members of a family for a mutation identified in the proband.

RESULTS:

We analysed 10 amplicons under a total of 59 temperatures. Successful results were obtained from 51 out of 59 tested temperatures (86.4%). In all cases a different heteroduplex-homoduplex pattern was obtained from mutant DNA in at least two of the temperatures. All 14 mutations identified by sequencing were also detected using dHPLC. Sequence analysis of a large family confirmed the dHPLC results.

CONCLUSION:

Using dHPLC we detected all mutations previously identified by sequencing.