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Genetic alterations of the APC gene in familial adenomatous polyposis patients of the hellenic group for the study of colorectal cancer.

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Source

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Abstract

Familial Adenomatous Polyposis (FAP)-a premalignant clinical entity inherited as an autosomal dominant trait-is characterized by the development of hundreds to thousands of adenomatous polyps of the colorectum during the second and third decade of life. Approximately 80% of the FAP patients harbour truncating germ-line mutations in the APC tumor suppressor gene (Adenomatous Polyposis Coli). We tested 48 members from 9 families. Two novel truncating mutations were identified-2601delGA, R923X--and five already known mutations R564X, R876X, Q1045X, 3927-3931delAAAGA and D1822V were found. Our method for testing was PCR amplification from genomic DNA extracted from whole blood, followed by automated DNA sequencing.