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Novel mutations of the APC gene in familial adenomatous polyposis in Greek patients.

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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 thousands of adenomatous polyps of the colorectum during the 2nd and 3rd decade of life. Approximately 80% of patients with FAP harbor truncating germline mutations in the adenomatous polyposis coli (APC) tumor suppressor gene. We tested 24 members of six Greek families. All patients had the FAP phenotype, and one patient had an extracolonic tumor (medulloblastoma). Our method for testing was the polymerase chain reaction (PCR) amplification from genomic DNA extracted from whole blood, followed by automated DNA sequencing. Two novel truncating mutations (2601delGA and R923X) and three already-known mutations (R876X, Q1045X, and D1822V) were found. Other polymorphisms were also found. We identified the inactivating APC mutation in 12 of 13 of our FAP patients. Our results suggest that PCR sequencing is a reliable method for screening the APC gene for germline mutations.