

Spectrum of the mutations predisposing to occurrence of colorectal polyposis.

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Polyposis colon is a major feature of several syndromes. Hereditary conditions that cause large numbers of intestinal polyps include familial adenomatous polyposis colonis (MIM 175100), familial adenomatous polyposis type 2 (MIM 608456), Peutz-Jeghers syndrome (MIM 175200), juvenile polyposis syndrome (MIM 174900), and PTEN hamartomatous tumor syndromes (PHTS). Polyps that arise from proliferative dysplasia are called adenomatous polyps or adenomas. They are true neoplastic lesions and are precursors to cancer. Hamartomatous polyps arise from abnormal maturation of the mucosa. They are noncancerous and have no malignant potential. In almost 30 years of polyposis research, we have studied just over 1,000 families with polyposis. Mutations were detected in more than 50% of families and were heterogeneous. We present here a summary of these studies in the face of new challenges related to the study of families in which mutations in predisposition genes have not been detected. The research in part was financed by the project of Polish Ministry of Science and Higher Education project number NdS-IISP0139202401

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