FAMILIAL ADENOMATOUS POLYPOSIS (FAP) IN CHILDREN: CLINICAL PRESENTATION, GENETIC BASIS, AND MANAGEMENT STRATEGIES

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Background. Familial adenomatous polyposis (FAP) is a hereditary disorder characterized by the formation of numerous adenomatous polyps in the colon and rectum, leading to a high risk of colorectal cancer if untreated. Objective of the study. The aim of this study was to explore the clinical presentation, genetic basis, diagnostic approaches, and management strategies for FAP in the pediatric population. Material and methods. A comprehensive literature review was conducted related to pediatric FAP from 2000 to 2023. Studies involving children (ages 0-18) diagnosed with FAP were included, while studies not distinguishing between pediatric and adult populations, focusing solely on sporadic polyposis, or lacking original data were excluded. Results. Although FAP primarily affects adults, its early onset in children poses unique diagnostic and management challenges. The prevalence of FAP in children was found to be between 1 in 8,000 and 1 in 10,000. Common clinical manifestations included rectal bleeding, abdominal pain, and diarrhea. with polyp detection typically occurring around the age of 15. Genetic testing for APC gene mutations was essential for early diagnosis. Endoscopic surveillance, for at-risk individuals, was crucial. Prophylactic colectomy was identified as the definitive treatment to prevent colorectal cancer, which posed a near-100% risk by the fourth decade if untreated. Conclusion. Early identification of FAP in children through genetic screening enabled timely surveillance and intervention, significantly reducing morbidity and mortality. Advances in genetic therapies and chemoprevention were suggested as promising areas for future research. Keywords: Familial adenomatous polyposis, FAP, children, pediatric, genetic testing, APC gene, colonoscopy, colectomy, colorectal cancer, early diagnosis.