Oncology
and Radiotherapy©
18(8) 2024: 001-007 • RESEARCH ARTICLE

Familial adenomatous polyposis: Diagnosis and treatment strategies

Abhinov Thamminaina[1], Kuldeep Singh Dodamani[2], Abhinav Mishra[3], K G Patel[4], Sulabh Mahajan[5], Rupa Mazumder[6]

[1] Department of Emergency Medicine, Krishna Institute of Medical Sciences, Maharashtra, India
[2] Department of General Surgery, Jaipur National University, Jaipur, India
[3] Chitkara Centre for Research and Development, Chitkara University, Himachal Pradesh, India
[4] Department of Biotechnology, Parul University, Vadodara, Gujarat, India
[5] Centre of Research Impact and Outcome, Chitkara University, Rajpura, Punjab, India
[6] Department of Pharmacy, Noida Institute of Engineering and Technology (Pharmacy Institute), Greater Noida, Uttar Pradesh, India

ABSTRACT

Objective: The evolution of many (>100) the colorectum is covered with colorectal adenomas is a hallmark of familial adenomatous polyposis. Both clinical and molecular methods may be used to identify this condition, which can be brought on using a lineage alteration within the adenomatous polyposis coli gene.

Materials and Methods: After receiving a description of the illness, individuals should have preventive proctocolectomy at the right time with a neo reservoir, often an ileoanal pouch. People with a family history of this condition who have not yet received a diagnosis should be encouraged to attend genetic counseling sessions and sign up for the proper clinical and genetic surveillance programs.

Conclusion: The gastrointestinal system may now be studied in more depth and with greater scope because to recent advancements the use of endoscopic technologies, such as triple-balloon endoscopy, double-balloon an endoscopy and high-resolution endoscopy. Further investigation into these novel endoscopic technologies may alter the surveillance strategies for hereditary polyposis of the adenomas, notwithstanding the paucity of data.

Keywords: human muty homolog-associated polyposis, attenuated familial adenomatous polyposis, adenomatous polyposis coli gene, familial adenomatous polyposis, genetic testing

Address for correspondence:

Abhinov Thamminaina

Department of Emergency Medicine, Krishna Institute of Medical Sciences, Maharashtra, India

E-mail: abhithamminaina@gmail.com

Word count: 5458 **Tables:** 04 **Figures:** 04 **References:** 29

Received: 14 August, 2024, Manuscript No. OAR-24-145464
Editor Assigned: 17 August, 2024, Pre-QC No. OAR-24-145464(PQ)
Reviewed: 01 September, 2024, QC No. OAR-24-145464(Q)
Revised: 08 September, 2024, Manuscript No. OAR-24-145464(R)
Published: 16 September, 2024, Invoice No. J-145464

INTRODUCTION

Hereditary colorectal cancer syndrome known as Familial Adenomatous Polyposis (FAP) is characterized by multiple adenomatous polyps of the Gastrointestinal (GI) mucosa. A lifetime risk of nearly 100% for Colorectal Cancer (CRC) is associated with FAP, which affects 1 in 7000–30,000 newborns. The colorectal cancer condition known as Familial Adenomatous Polyposis (FAP) runs in families [1]. There is no difference in the likelihood of the disorder being passed down from one generation to the next based on the gender of the parents, and the average age at which symptoms of the disease appear in a patient is around 15 years [2]. Before this age, it is stated that there are no symptoms of the condition, and there are also no macroscopically apparent polyps. The majority of the time, a germline pathogenic mutation in the chromosome-located Adenomatous Polyposis Coli (APC) tumor inhibiting gene results in FAP [3]. The formation of more than one hundred colorectal adenomas is the principal effect of this mutation, and it nearly always has this effect. The presentation is often asymptomatic the great bulk of the time, although some patients may have symptoms such as diarrhea, rectus bleeding and stomach discomfort, tenesmus, and blockage, typically during the 2nd or 33 years of life [4].

There are several rare instances of FAP that present with symptoms within the first decade of life. The reports of people suffering with FAP are quite unusual. To discuss an instance when a 13-year-old girl who showed signs of who hematochezia and was ultimately determined to have FAP as the underlying condition [5]. According to the criteria established by SCARE, this case has been reported. The precancerous disease known as Familial Adenomatous Polyposis (FAP) is passed down in an overriding genetic fashion and affects around one in every 10,000 people. It is distinguished by the occurrence of several gastrointestinal adenomas in addition to extracolonic symptoms [6]. If left untreated, up to one 100% of these individuals may develop adenomas of the duodenum and periampullary area, and there is a strong risk that these adenomas will proceed into adenocarcinomas. Because of the progress made in genetic screening technologies, the mortality rates of carcinoma those colon patients who have this illness have dropped to the point that duodenal cancer and desmoid tumors are now the primary reasons for death. A condition known as chromosomal abnormalities are a hallmark of Familial Adenomatous Polyposis (FAP). rapid development of a significant number of colorectal adenomas at a young age [7]. FAP is a result of heritable changes

to the APC gene that is present in germline. The presence of more than one hundred adenomas and a young age of initiation of polyposis are the defining characteristics of classical FAP in many cases, thousands of adenomas are found. Although it is responsible for less than one percent of all cases of colorectal cancer, it has a connection to an almost one hundred percent lifetime likelihood of developing the disease. gastrointestinal polyps and duodenum are a common extra-colonic manifestation, as are osteomas, desmoid tumors, and numerous lesions referred to as ocular lesions that have pigment (congenital enlargement relating to the retinal pigment epithelium) [8]. There is a possibility that up to thirty percent of FAP instances are the result of spontaneous germline mutations, also known as de novo mutations, or mosaicism. Attenuated FAP (AFAP) is likewise a result of autosomal dominant mutations in the APC gene however, the illness manifests itself much later and there are fewer adenomas. Patients who have a cumulative count of 10 or more adenomas before the age of 30 years or between 20 years and 99 years adenomas at any age should be evaluated for the condition. When AFAP is present, adenomas may mostly appear in the proximal colon, and there is often a great deal of phenotypic diversity within a family. The forms of extra-colonic malignancies that are more prone to occur in patients with FAP include hepatoblastoma, brain, thyroid, and upper gastrointestinal tract (most often the duodenum) cancers. Desmoid tumors are also more likely to develop as a result of this condition [9]. A hereditary recognized as adenomatous polyposis in the family, or FAP, generally occurs in the first 10 years of life and is characterized by the growth of many adenomatous cells in the rectum and colon. In FAP, there are more than 100 polyps. Gardner Syndrome is another name for FAP, which is also known as familial polyposis coli, Adenomatous Polyposis Coli (APC), and familial adenomatous polyposis coli. People who have FAP have an increased risk of developing colorectal both cancer and other diseases tumors. APC gene mutations, which may be found on chromosome 5q21, are responsible for the majority of FAP cases. FAP is a genetically predisposed illness, meaning that those who have it are genetically heterozygous. This indicates that there is a 50% possibility that a patient with FAP will pass on the illness to each of their offspring. Because they possess both a mutant and healthy variant of the APC gene., affected people are genetically heterozygous. There is an equal risk that it will have an effect on boys and girls. There are several APC mutation types that have been identified in FAP, including insertions, deletions, and nonsense mutations [10].

These mutations may cause transcriptional frameshifts and/or early stop codons. The prospective endoscopic study demonstrated that CE had a substantial influence on the adenoma identification and wholesome treatment in the upper digestive system. To the best of the knowledge, this experiment was the biggest one ever conducted on patients with FAP. This results in more frequent and stringent monitoring checks [11]. Study determined whether or not the differences in the metabolomics reflected the effects of FAP the serum, as well as to learn about significant metabolic changes connected to the etiology of FAP, which could then be utilized to enhance FAP therapy approaches, followed by a chance to stop FAP from developing into CRC [12]. In addition, the clarification of clinical conditions in the FAP's monomic properties has the potential to assist to the examination of changes caused by APC mutation in CRC patients. The unusual example demonstrates the need to take into account the higher danger of fibrosis in FAP patients, even in the classic type, before electing to implant breasts [13]. The research was to examine the frequency clinical risk elements, and a prevalence of pouch adenomas potential phenotype-genotype relationship in a large group of individuals accompanied with familial adenomatous polyposis. It also examined pouch adenoma-free survival [14]. The study will go through this polyposis syndrome's genetic and clinical elements, detection, extracolonic symptoms, and tumor characteristics [15]. The work, which focused on metabolite variations, may provide useful insights into the biochemical process of FAP, the changes in metabonomic brought on by the APC mutation, and the mechanisms behind the development of CRC [16]. The retrospective investigation, all patients who had surgery for either sporadic or FAP duodenal cancer between 2000 and 2014 were identified. In order to compare the perioperative and survival results, the patients were categorized according to diagnosis [17]. Study cross-sectional research was to investigate the connection between CRC development in FAP patients and physical fitness. A step test was performed on 119 individuals with FAP (54 men; 65 women), ranging in age from 17 years to 73 years [18]. Each patient's predicted maximum oxygen uptake (VO_2max) was computed using heart rate as a measure of physical fitness. Patients with Familial Adenomatous Polyposis (FAP) far more commonly exhibit this illness. Adenomatous Polyposis Coli (APC) gene mutations in the germline are the cause of the autosomal dominant hereditary illness known as FAP. A comparative study between family kinds and sporadic types was conducted to highlight the specific clinical characteristics of CMV-PTC. Research indicates that when paired with sulindac, changing from continuous the effective treatment of adenomatous tumors in the intestines and small intestine involves administering erlotinib once a week at a fraction of the present therapeutic dose [19]. The research clusters with a phenotype marked by desmoid tumors, widespread stomach polyposis, and colon oligo-polyposis. To end by identifying a distinct clinical variety of FAP that it would like to call this condition Gastric Polyposis and Desmoid FAP, which may require specialist care. Research clusters characterized by desmoid tumors, extensive stomach polyposis, and colon oligo-polyposis [20]. Research was to use decision analysis to assess the compromises between complete proctocolectomy with IPAA and total colectomy with ileorectal anastomosis [21]. Research sought to determine the incidence that extracolonic cancers a sizable group of Adenomatous Polyposis Coli (APC) mutation carriers as well as to determine the reasons of mortality [22]. From the Dutch polyposis registry, all APC mutation carriers were chosen. On death causes, information was gathered. From the Dutch Pathology Registry, pathology reports were obtained. Research assessed possible predictive biomarkers for celecoxib's chemo preventive efficacy [23]. Research showed that celecoxib's bioavailability differed between normal and polyp tissues and that it may have an impact on the Clinical response to chemoprevention in FAP patients [24]. Patients with dual CD and FAP diagnoses have more difficulty in making this choice [25]. To describe a young woman with FAP whose active CD was found in the ileocaecal area. She was advised to have surgery (colectomy with terminal ileostomy and terminal ileum resection) due to several big colon polyps and a stenotic terminal ileum. It was then constructed to create an ileorectal anastomosis [26]. The research is to look at the relationship between Colorectal Adenomatous Polyps (CAP) and stomach H. pylori in the Chinese population [27]. The purpose of

the research was to outline the TAP phenotypic range in a multi-institutional population. The originating derived from eight high-risk cancer facilities, TAP cases were found. Patients with more than 10 gastrointestinal polyps and a history of CYAC treatment in the past who had no known hereditary colorectal cancer risk syndrome or causal germline change were considered cases [28]. It is not advised to apply this technical standard in the clinic for patient assessment [29]. To choose an appropriate testing plan and direct medical screening and care, kindly consult the National Comprehensive Cancer Network (NCCN) clinical practice recommendations [30].

MATERIAL AND METHODS

In this part, will go into great depth regarding a qualitative literature evaluation that was carried out with the intention of assessing. The consequences of having hyperemesis gravidarum during pregnancy, in addition to experiencing nausea and vomiting throughout pregnancy.

Epidemiology

CRC is a significant participant in worldwide mortality and morbidity due to cancer. The incidence of it varies greatly in terms of populations, are highlighting the biggest rate. Around the world, 85% of CRCs are thought to be sporadic, 15% are familial, and less than 1% is thought to be caused by FAP even though one of most well-known and comprehended hereditary diseases is FAP (Figure 1 and Table 1).

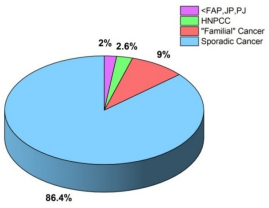

Fig. 1. The approximate and related effects of family characteristics on colorectal cancer prevalence

Tab. 1. The proportionate and approximate roles that family factors play in the occurrence of colorectal cancer

HNPCC	2.6
Sporadic Cancer	86.4
<FAP, JP, PJ	2
"Familial" Cancer	9

There are local FAP registers in many nations, but it is difficult to get reliable statistics. It is estimated that 1 in 8,300 babies were born with FAP. Before mutation analysis was available and all clinical variants and differential diagnoses were recognized, these projections were made based solely on clinical criteria. FAP afflicted between 11,300 and 37,600 persons, or 3–10/100,000 people. By the late teen and early twenties age range, FAP clinically manifests in both sexes equally.

RESULTS AND DISCUSSION

Clinical description

Children and adolescents seldom have symptoms prior to the adenomas being big and numerous enough to result in rectal either hemorrhage or anemia. Recto-sigmoid examinations may be used to identify polyps that are indicative of FAP and can be prompted by other non-specific concerns such weight loss in young patients, stomach pain, diarrhea or constipation, changes in bowel habits 0discomfort, or palpable abdominal lumps. FAP can show osteomas are extraintestinal appearances, atypical teeth, congenitally lacking teeth, odontomas, desmoid tumors, dentigerous cysts, and Congenital Retinal Pigment Epithelium Hypertrophy (CHRPE), and cancerous growths outside of the colon. The Gardner type of FAP is characterized by a number of symptoms in addition to osteomas of the skull and maxilla. Nowadays, the illness will almost always present as colon cancer, much less an extra-colonic malignancy.

Other gastrointestinal manifestations

The FAP is certain extra-colonic gastrointestinal symptoms may also manifest. Fundic Gland Polyps (FGP) occur in the stomachs of 90% of FAP patients. Compared to spontaneous FGP, which is benign, 40% of lesions in people with FAP have been shown to contain adenomatous characteristics, however they seldom develop to malignancy, making them particularly interesting. FGPs in patients with FAP differ pathogenetically from FGPs that arise at random. APC gene changes that are multiple somatic, second-hit in FAP-associated FGPs precede morphological dysplasia, indicating that these FGPs are neoplastic lesions.

In one study, they appeared 10 years-20 years following the identification of colorectal polyps in almost 90% of those with FAP. According to some reports, the risk of duodenal adenomas during life might exceed 100%. Duodenal polyps were ranked by Spigelman according to their degree, histology, number, and size of dysplasia (Table 2). 5% of duodenal polyps, namely periampullary polyps, are estimated, are predicted to progress to malignancy in less than a decade. Although uncommon in the general population, FAP patients have a several hundred-fold increased risk of duodenal or periampullary cancer. Although lower Spigelman stages can be found in patients who are being monitored for cancer, duodenal polyposis typically development is orderly manner with a bigger Spigelman stages. A malignancy may show as pancreatitis or an ampullary adenoma may cause.

Tab. 2. Spigelman grading system for FAP-related duodenal polyposis

	Phase 3	Phase 2	Phase 1
Polyp size (mm)	>10	05-10	01-04
Histology	Villous	Tubulovillous	Tubular
Polyp Number	>20	05-20	01-04
Dysplasia	High grade	Low grade	Low grade

It is widely acknowledged that individuals with FAP are more likely than usual to develop ampullary and duodenal neoplasms as well as small intestinal polyps and even cancer. The actual frequency of small intestinal polyps is uncertain, and it often depends on the examination technique. By using push enteroscopy, Bertoni G et al. investigated 16 people who have FAP in 1993 and revealed that 50% of the patients had jejunal polyps [13]. There was a 30%–75% incidence of jejunal and ileal polyps when double balloon enteroscopy and capsule endoscopy were used [14-16]. Compared to ampullary or duodenal cancer, small bowel cancer occurs at a significantly lower incidence. The treating physician should be prepared to implement surveillance, which will be covered, and should be informed of this potential.

Other extracolonic malignancies

Pancreatic mucinous adenocarcinomas, liver (Hepatoblastoma), and brain cancers are a few more extra-colonic malignancies connected to FAP. The development of malignant central nervous system tumors in two adolescent brothers with adenomatous colorectal polyps was first documented by Turcot and colleagues in 1959. Then, it was discovered that "Turcot syndrome" has at least two subgroups and is heterogeneous. The first, patients with Lynch syndrome manifest with glioblastoma due to a germline mutation in either of these DNA mismatch correction genes, hPSM2 or hMLH1. When FAP and APC germline mutations are present, medulloblastoma, or in rare cases, glioblastoma, are indicative of a second Turcot subtype. The first instance of thyroid cancer in a person with FAP was reported as of 1949, Crail. Thyroid cancer is particularly common in young women (under 35), with a prevalence that is almost 160 times higher than the overall population.

Etiopathogenesis

FAP is a genetic disorder brought on by an APC mutation. Most FAP patients are "de novo," indicating there lacks clinical or genetic evidence that their ancestors have the disorder, even if 25%–30% of them have family records of colorectal tumors and cancer. It is now understood that germline mosaicism may contribute to this, at least in part [28]. A germline APC mutation predominantly causes classic FAP, an autosomal dominant characteristic; specific APC mutations are the main cause of AFAP. Instead, a portion of people with the MUTYH gene mutation will cause the clinical signs of FAP.

APC gene

A gene called band q21 (5q21) is located on chromosome 5's long arm, there is a tumor suppressor gene called PC. A big protein (309 kilo-Daltons) is encoded by the area of coding, this is divided into 15 exons. The APC protein includes a number of oligomerization- and binding-mediating domains that interact with a number of intracellular proteins that are crucial for cell adhesion, signaling, and activation of transcription.

Typical APC features and organization

The APC gene has a 108,353 base pair length. The mRNA has 16 exons and 10,719 base pairs. A protein with a length of 2,843 amino acids and a molecular weight of 310 kDa is encoded by the mRNA. The final exon, exon-16, is 8,689 bp long and contains the majority of the amino acids out of the 6,574 bp of coding sequences. There are 653 amino acids in just exons 2 to 15 which for protein, compared to 2,190 in exon 16. Tumor suppressor protein APC is widely recognized for its ability to regulate-catenin breakdown, which is a necessary component of Wnt signaling. Glycogen Synthase Kinase 3 (GSK3), conducting, and -catenin form a protein complex is affected by wnt signals in terms of sta-bility. APC protein in its wild-type form or without Wnt causes the degradation of -catenin. Target genes for -catenin, C-myc, among others, are expressed when Wnt is present or when APC is not (as is the case in many cases of colon cancer). The polyamine Ornithine Decarboxylase (ODC), a proto-oncogene, is then ex-pressed as a result of myc expression (Figure 2). Through its inter-action the APC gene product collaborates with the transcription factor-catenin to regulate the transcription of many crucial genes involved in cell proliferation. Chromosome attachment in culti-vated cells has been demonstrated to be impacted by APC trunca-tion in homozygotes. APC as shown by have roles in both in vitro and mouse model studies of cell migration.

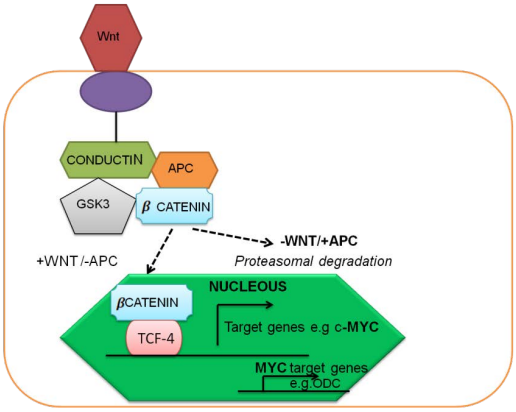

Fig. 2. Relationship between Wnt signaling and the APC tumor-suppressor

Modified APC configuration and function

As of right present, mutations are linked to FAP in over 300 different kinds. A shortened most often, these changes result in protein (insertions, deletions, nonsense mutations, etc.). The codon 1309 deletion mutation, which affects 10% of FAP sufferers are the most frequent change. Codon 1061 deletion mutation, which affects 5% of patients, is the second most frequent mutation.

Humans and animal models of APC-disturbed FAP structure and function

It is known that loss of normal APC activity, which takes place in the pre-adenoma stage, occurs early both spontaneous and familial colon cancer pathogenesis. Typically, colon tumors exhibit either the instability of microsatellites, either, but not both, is associated with loss of mismatch repair activity.

Genetically engineered mice offer a wonderful in vivo system for human illnesses as well as an opportunity to test remedies. The APC gene has a point mutation in the FAP mouse model, the original framework for examining how the APC gene contributes to the genesis of intestinal malignancies, and it produces a lot of adenomas. Using a mouse model, loci are modified in order to illustrate the idea of genetic control illness severity.

The majority of nonsense mutations found in germ lines from people with FAP cause a truncated protein to be produced, based on APC gene mutational research. The central section of the protein, between codons 1284 and 1580, is known as the Mutation Cluster Region (MCR), is home to more than 60% of APC mutations.

Genotype-phenotype correlation

There is some correlation between clinical indications of an illness and the sites of certain genetic abnormalities, but it is not perfect and there are some discrepancies (to be explained later) (Figure 2). There is a link between profuse polyposis and mutations between codons 1250 and 1464. CHRPE is almost always absent when an APC gene mutation that truncates proteins occurs before exon 9, it occurs after this exon, it is consistently present. Patients who have mutations in the region between codons 1445 and 1578 may not convey CHRPE but may still get severe desmoid tumors.

Diagnosis

Clinical symptoms and a suggestive family history are used to make the diagnosis of typical FAP. Genetic testing should, if feasible, be used to confirm the clinical diagnosis.

Clinical diagnosis

Screening and early presymptomatic identification of members of at-risk families are 2 of the main goals of genetic testing. Furthermore, it's critical to confirm the diagnosis in patients who have hazy clinical findings. Until results are available, management of first-degree relations therapeutically. Initially, just the index case is looked at. This is due to the fact that it may frequently take a while before the initial mutation study's findings are available. If the mutation is known, a quick and affordable test may be performed to identify family members who are at risk.

Human MUTYH structure and function at normal levels

Screening Several areas of the gene for the MUTYH comprise DNA required sites, an adenine binding motif, and numerous interactions between the APE1 enzyme, MSH6, PCNA, and Replication Protein A (RPA) (Figure 3).

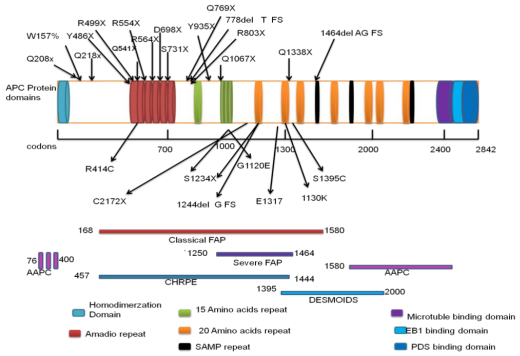

Fig. 3. Mutations in the APC protein domains and their relationships to FAP phenotypes

MUTYH-disturbed structure and Associated Polyposis (MAP)

Every normal protein activity of the MUTYH gene may be altered or totally stopped by DNA changes in functional domains. For instance, one type 2 alternative splicing mutation might lead to a gene-based item having 521 amino acids (instead of 546). Unlike type 2, which is taken to the nucleus and lacks the initial exon encoding an MTS, type 1 is transferred to the mitochondria. Virtually all carriers of the bi-allelic MUTYH mutation are affected by colon polyp penetrance, and patients commonly experience the colon and rectum to acquire 10–100 adenomatous polyps/adenomas. Some evidence points to a somewhat increased risk of Colorectal Cancer (CRC) for heterozygous MUTYH mutation carriers, who are most likely dominant MAP types (45, 46). Adenomas, or upper gastrointestinal polyps, affect around 1/3 of the population.

Management of the FAP patient

The major objectives in managing people the objectives of FAP indications or preventing cancer and preserving a good standard of living are examples of genetic evidence. The most crucial clinical test is a large bowel endoscopy since CRC is nearly always present. However, as was previously mentioned, the disease is widespread and manifests outside of the colon, so it should be searched for by routine reexaminations. Furthermore, depending on the Spigelman stage and polyp burden, upper tract endoscopies with a side and front view must be carried out either yearly or every five years to identify gastric but most often periampullary and duodenal adenomas (Table 3).

Tab. 3. Recommended surveillance interval intervals between gastroscopy examinations

Spigelman 0 and I:	5-year gaps between endoscopies
Spigelman Stage II:	3-year gaps between endoscopies
Spigelman Stage III:	1 year-2-year gaps between endoscopies Think about endoscopic ultrasound Think about celecoxib 800 mg daily
Spigelman Stage IV:	Ultrasound imaging during endoscopy Think about surgery: Duodenectomy with pylorus- or pancreas-sparing surgery

Because adenomas are becoming more common, prophylactic colorectal surgery in the late teens or early 20s is frequently advised. While this is a discretionary formula, the time can be set up to cause the patient and family the least amount of inconvenience. Elective surgery may sometimes be postponed if the patient is cooperative and the polyps are few and small.

Chemoprevention

According to andomized studies. Celecoxib and rofecoxib (108, 109), two particular Cycloxygenase-2 (COX-2) inhibitors, were created to reduce the likelihood of gastrointestinal harm brought on by reducing the cytoprotective COX-1. behavior accompanied with for six months of patients were given 400 mg of celecoxib twice a day with FAP is observed to lower the tumor burden by 28% as opposed to 4.5% of the placebo group, as shown in figure 4. (Table 4)

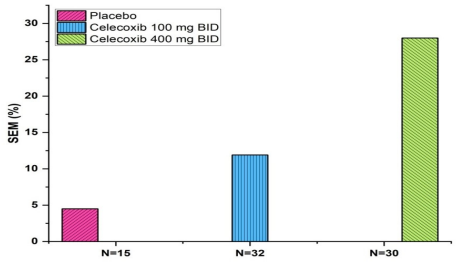

Fig. 4. Analysis of colorectal polyps in FAP patients

Tab. 4. Colorectal polyps in individuals with FAP

	Placebo	Celecoxib 100 mg BID	Celecoxib 400 mg BID
N=15	4.5	-	-
N=32	-	11.9	-
N=30	-	-	28

Chemoprevention

According Sulindac was not demonstrated in order to stop primary development of adenomas in FAP patients in a recent study. Even though COX-2 inhibitors are typically safe for digestive system toxicity, prolonged usage of these medications for avoiding CRC poses a modest risk of serious cardiovascular problems. The treatment of Duodenal or retained rectum adenomatous polyps with CRC and intramucosal adenomas continued to develop despite the high cost of celecoxib/sulindac. All adenomas contain COX-1, which if left untreated could be the cause of this. Adenoma regression has also been linked to oral estrogen/progesterone contraception.

Attenuated FAP

The therapy of AFAP patients is greatly influenced by the number and location of polyps in the large bowel. Colonoscopic polypectomy is adequate in patients with a few adenomas that can all be removed. A 2-year gap among colonoscopies, likely for life, may be appropriate because these have an adenoma-carcinoma sequence people does not seem to be unduly accelerated. The preferred course of therapy for these individuals is surgical resection if several polyps or clusters are discovered during an endoscopy if repeating complete strictly speaking, a colonoscopy challenging. An ileorectal anastomosis is frequently performed in conjunction with a subtotal colectomy because of the relative rectum sparing in AFAP.

CONCLUSION

Familial Polyposis Coli (FPC) and Familial Adenomatous Polyposis (FAP) are two autosomal dominant polyposis syndromes with differing levels of penetrance. In the colon and rectum, patients will grow hundreds to thousands of polyps if left untreated. Too looked at the research on methods for the detection, monitoring, and treatment of FAP. The introduction of HR endoscope, endoscopic capsule, and double-balloon endoscopy, the gastrointestinal system has become the subject of increasingly thorough and comprehensive study. The potential of these novel endoscopic tools to improve FAP monitoring tactics will become clearer with more research.

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