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Familial Adenomatous Polyposis: A Case Report of An 8-year-old Boy

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Abstract:

Familial adenomatous polyposis (FAP) is characterized by the development of up to hundreds or thousands of adenomas in the colon and rectum with several extra colonic manifestations. These syndromes have multiple genetic characteristics and are associated with an increased risk of colon cancer. An 8-year-old boy presented to our department with a history of repeated episodes rectal bleeding requiring blood transfusion for last 3 month. His laboratory evaluation showed normal hematological parameter. He had multiple polyps on endoscopic examination with histopathological confirmation of adenoma. As genetic testing is not possible in our country, on the basis of clinical presentation and histopathological confirmation he was diagnosed as a case of Familial adenomatous polyposis (FAP). Though Familial adenomatous polyposis (FAP) is the most common type of polyposis in children, it is very rare in our country.

Key words: *Familial adenomatous polyposis, adenomatous polyposis coli, Children.*

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Introduction:

Familial adenomatous polyposis (FAP) is the most common type of inherited polyposis syndrome in children which is characterized by the development of up to hundreds or thousands of adenomas in the colon and rectum and several extra colonic manifestations. FAP is an autosomal dominant disorder due to adenomatous polyposis coli (APC) gene mutation. The gene responsible for FAP, APC (adenomatous polyposis coli), is located on chromosome 5q21 and appears to be a Tumour suppressor gene. Incidence is 1 to 3 in 10,000 births& penetrance is almost 100%. Spontaneous mutation in FAP is 20% to 30% cases.[1]Patients may be asymptomatic even with numerous polyps, but may present with blood or mucous mixed stool. [2]Colonoscopy shows huge number of small,nodular, and commonly sessile polyps and histology reveals tubular, tubulovillous, and villous adenomas with dysplasia.[3] FAP should be treated by colectomy as there is risk of colorectal cancer. Also regular surveillance is recommended for FAP patient as they are associated with an increased risk of colon cancer.[1]

Case report:

An eight year old boy, only issue of non-consanguineous parents, got admitted in Bangabandhu Sheikh Mujib Medical University (BSMMU) on September, 2022 with the history of repeated episode of painless massive rectal bleeding for last 3 month. The bleeding was sudden, 100 -200 ml in each episode, dark red colour in colour, foul smelling & associated with abdominal pain followed by severe pallor and lethargy. He got six units of blood transfusion before admission.Then patient got admitted in this hospital for evaluation of repeated per rectal

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bleeding. He had no family history of such type of illness. On physical examination he was vitally stable, no pallor, anthropometrically well thrived. His laboratory Investigation showed hemoglobin 12.2 gm/dl, ESR 10 mm in 1st hour, platelet count 2,80,000/L, total count 5,500 /L. Neutrophil 53%, lymphocyte 37%, C-reactive protein 0.3 mg/dl, prothrombin time 12 second, international normalization ratio 1.01, activated partial thromboplastin time 28 sec. Colonoscopy showed numerous small, nodular polyps present in the ascending colon, and transverse colon with a small polyp in cecum. Endoscopy of upper gastrointestinal tract finding revealed one sessile polyp present in second part of duodenum. Multiple biopsies were taken from polyps and histology confirmed tubular adenoma. Patient was also investigated for other FAP associated lesions for extra colonic manifestation and all reports were normal. Confirmed diagnosis for FAP on the basis of genetic testing was not possible in our country for APC gene. But in this case, diagnosis was made by typical clinical features and histological findings. He was referred to surgery department for colectomy. As earlier age of presentation, on histology there was no dysplastic changes they postponed colectomy & advised patient to come for follow up whenever he develop rectal bleeding.After discharge he developed rectal bleeding for 2 episodes without requiring blood transfusion. We advised patient yearly colonoscopy to see polyp burden, distribution, progression & histology.

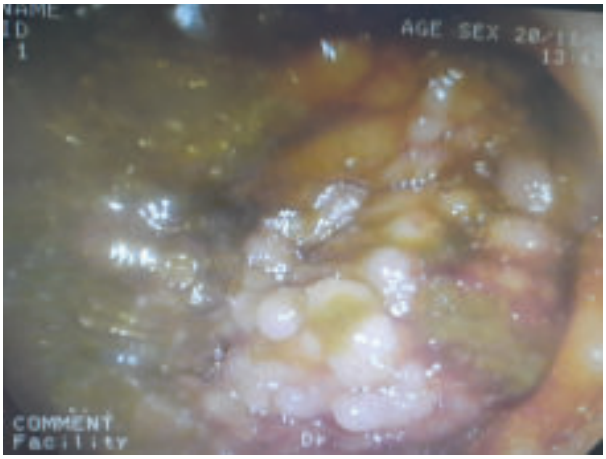

Fig. 1: *Numerous small sessile nodular polyps in colon*

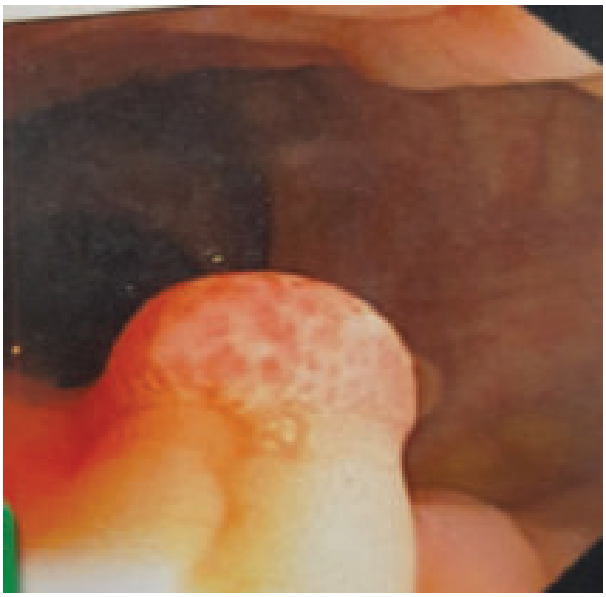

Fig. 2: *A sessile polyp in 2nd part of duodenum.*

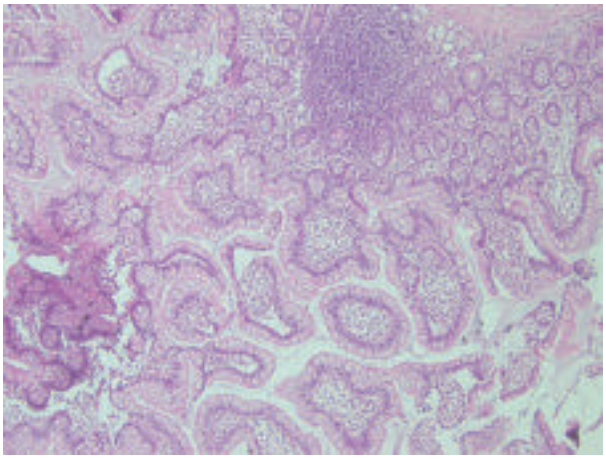

Fig. 3: *Light microscopy reveals microscopic adenoma.*

Discussion:

FAP is characterized by the progressive development of hundreds to thousands of adenomatous polyps in the large intestine. FAP usually begins to appear in childhood or adolescence and increase in number with age.[4]A case series showed mean age of FAP was 13 years.[5] Although adenomas may first appear at age 8 to 12 years, these are largely not clinically significant.[1] Our patient presented at early age with significant bleeding. Children with FAP who develop severe polyposis of the colon before the age of 10 years is considered having mutation located at codon 1309 in the APC gene.[6] FAP is inherited as autosomal dominant pattern, in 20–30% of cases there may be

spontaneous mutations.[1] There was no family history in this patient indicating spontaneous mutation. In one study family history was positive in 56% patient and another study showed family history present in 67% patient having FAP.[3,7] FAP affects both genders equally and has a worldwide distribution.[7]

On Physical examination this patient was vitally stable and there was no pallor, during admission there was no ongoing bleeding and he got 6 unit of blood transfusion before admission. Physical examination is often nondiagnostic in children with FAP unless there is evidence of rectal bleeding or hypoproteinemia.[3] Rectal bleeding was most common symptom (86%) in previous study.[7] At colonoscopy numerous adenomatous polyps can be seen which are small, nodular, and typically sessile, and of variable size. On histology, the polyps can be tubular, tubulovillous, and villous adenomas with dysplasia ranging from low to high grade. Polyps involving the entire colon and a polyp burden of more than 50 polyps at the time of diagnosis was found in the majority of patients in a recent large pediatric series.[3] The standard clinical diagnosis of typical/classical FAP is based on the identification of >100 colorectal adenomatous polyps.[1] Colonoscopic examination of this patient revealed numerous small nodular sessile polyp over transverse colon, ascending colon and cecum. But we could not estimate the accurate number of polyp. Histopathologic examination revealed adenomatous polyp without dysplastic changes.

A polyposis syndrome can be defined as either carrying a known mutation for one of the polyposis syndromes or having multiple polyps on endoscopic examination with histopathological confirmation of adenoma. Clinical diagnosis of FAP should be confirmed by genetic testing if possible.[5] However, genetic testing is not possible in our setting. We diagnosed our patient on the basis of endoscopic evaluation and microscopic confirmation of adenoma. When colonic adenoma have been identified in a child at colonoscopy, for example, for rectal bleeding, they should be examined for extra colonic features of FAP, for example, skin, dental, or bone manifestations.[1] Our patient had no skin, dental or bone manifestation, but he had a sessile polyp in 2nd part of duodenum on esophagogastroduodenoscopy. Upper Gastro intestinal polyp were documented in 58.9% patient of FAP patient via upper GI endoscopy, of which 51.5% had stomach polyp & 33.7% had duodenal polyp.[3] Another study revealed 50% patient had gastric fundic gland polyposis and 50% had duodenal adenomatous change.[7]Duodenum is the most common site for adenomatous polyp of FAP patient commonly arising in the second or third part of duodenum.[8]

Development of colon cancer is inevitable in FAP patient unless colectomy is performed; accounting for 1% of all colorectal cancer patients.[9] So removal of the colon for FAP is required to prevent the almost inevitable development of colorectal cancer. There are no guidelines regarding the timing of surgery, nor evidence to dictate the point at which colectomy should be performed based on polyp burden (number or size). The adenoma-carcinoma sequence is not accelerated in FAP, so it may take more than 10 years before a cancer develops in a colon with adenomas. As our patient was young and histopathology report showed no dysplastic changes, prophylactic colectomy was postponed. Without colectomy, affected individuals usually develop colorectal cancer by the third or fourth decade of life.[10]

The two main surgical options are colectomy with ileorectal anastomosis (IRA) and proctocolectomy with ileal pouch-anal anastomosis (IPAA). Both surgical choices have their merit and weakness. The IRA is a can be readily performed laparoscopically, reducing hospital length of stay and recovery with preservation of bowel function and continence, and small surgical scars with cosmetic advantage. The IPAA requires more extensive surgery with pelvic dissection with its attendant risks of hemorrhage, damage to pelvic nerves, and possible reduction in fertility.[11]

We advised our patient surveillance colonoscopy 1-3 yearly or early whenever significant bleeding. The risk of developing cancer in teenagers is as low as 0.2% (10), so waiting 1 to 3 years between colonoscopy would appear safe, so long as families are not lost to follow-up if endoscopies are as far apart as every 3 years[4]. If there is sufficient concern about polyp size, polyp density, and the presence of feature suggestive of advanced changes, the patient should be referred for colectomy.There is no justification for routine biopsies or polypectomy to assess dysplasia. Waiting for serial biopsies to change from low-grade to high-grade dysplasia is unsafe and will put patients at risk of developing colorectal carcinoma especially as it is not clear which polyps should be biopsied at colonoscopy for histological assessment.[12]

Conclusion:

Polyposis in childhood is harbinger of colonic and extra colonic cancer. So lifelong surveillance for these patients are mandatory. Any child presented with sudden severe rectal bleeding, suspicion should be made of FAP. If we can bring every FAP patient under surveillance by early diagnosis major complications can be avoided and long-term outcome will be improved in these patients.

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