CASE REPORT

Laparoscopic total colectomy in an eight-year-old with familial adenomatous polyposis: A case report

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Abstract

Familial Adenomatous Polyposis accounts for <1% of all the colorectal cancer cases, with progression to colorectal cancer usually at ≥20 years of age. Endoscopy is essential for the diagnosis with definitive treatment involving prophylactic total colectomy. With current surgical advances, this is routinely being performed with the aid of laparoscopy. Due to resource limitations and non-availability of the screening programs in the developing world, such cases remain under diagnosed. Genetic testing is necessary for prognostication of both the index case and their at-risk family members. Thus, we present a rare case of an eight-year-old female, with an early onset progression to colorectal cancer with Familial Adenomatous Polyposis. We performed a prophylactic laparoscopic total procto-colectomy with ileo-anal anastomosis, which to our knowledge is the first ever-performed procedure in Pakistan. We conclude that progression to colorectal carcinoma in familial adenomatous polyposis can present at an earlier age than that reported in the literature. Laparoscopic total colectomy has similar outcomes than open surgical methods with better cosmetic results.

Keywords: Familial adenomatous polyposis, Colectomy

https://doi.org/10.5455/JPMA.27928

Introduction

Familial Adenomatous Polyposis (FAP) is an autosomal dominant, inherited predisposition for the development of multiple adenomatous polyps of the colon. The mean age of onset is around 15 years; however, it may only be symptomatic by the third decade with a median age for the development of colonic cancer being as early as 35-40 years.[1] The probability of developing cancer is almost 100%. A tumour registry index for paediatric cases of colorectal cancer associated with familial adenomatous polyposis does not exist. As a prophylactic or curative procedure for FAP, a total proctocolectomy with ileal pouch anal anastomosis (IPAA) or total colectomy with ileorectal anastomosis (IRA) is often indicated. Rectal and colonic polyps are seldom observed before 10 years of age. These patients are also at an increased risk of extra-colonic malignancies, such as duodenal ampullary cancer, follicular or papillary thyroid cancer, childhood hepatoblastoma and medulloblastoma.[2]

Herein, we present a case of an eight-year-old female who presented with symptomatic FAP with progression to colorectal cancer diagnosed post surgically. We have also discussed the surgical details of a first ever laparoscopic total proctocolectomy performed in the paediatric age group in Pakistan. Consent was obtained from the child's elder brother who acted as her guardian in the absence of her father.

Case Report

An eight-year-old female presented to the gastroenterology clinic at the Aga Khan University Hospital on 26th April 2018, with complaints of per rectal bleeding and abdominal pain since the last two weeks. This was coupled with decreased appetite. Per rectal bleeding was intermittent with production of fresh blood on defecation. Abdominal pain was intermittently present

Figure-1: Total colon removed intraoperatively. Multiple polypoidal growths all throughout the colonic mucosa with normal mucosal pattern in between.

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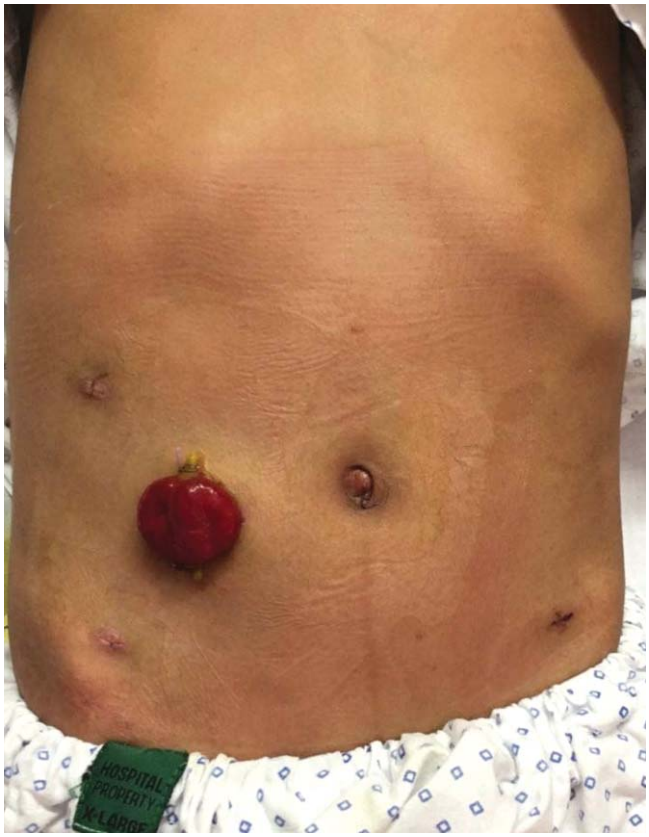

Figure-2: Postoperative image of the surgical site reveals no incision mark and better cosmetic outcome with only ileostomy visible.

and vague in nature mostly located in the lower half of the abdomen. This was coupled with decreased appetite since the last 4 months. She had received a full course of her childhood vaccines. Her past medical history and the birth history were unremarkable. She had been transfused one unit of packed red cells three weeks prior to the presentation. Her parents had a consanguineous marriage with no prior comorbid conditions present within the family, except that, an elder female sibling had passed away at the age of fourteen years due to chronic kidney failure.

On general physical exam, she was an emaciated girl with a visible pallor and dehydration. Abdominal exam was within the normal limits. Per rectal exam revealed no lesion or active bleeding at the anal verge. Initial differentials were mapped out to the celiac disease, abdominal tuberculosis or solitary rectal polyp. Investigations revealed a decreased haemoglobin level of 7.30mg/dl (11-14.5 mg/dl), decreased mean cell volume of 64 fL [78.1-95.3 femtoliters(fL)], stool for *Helicobacter pylori* antigen was negative. Malarial parasite immunochromatography was also negative along with a negative Mantoux test, her tissue transglutaminase Immunoglobulin A was and Immunoglobulin G were within the normal limits.

She underwent a diagnostic flexible colonoscopy, which revealed multiple sessile polyps in descending and sigmoid colon, along with multiple polys in the rectum. Twelve polyps were extracted and sent for histopathological examination, which revealed tubular adenoma with high grade dysplasia. A Paediatric Geneticist opinion was sought, for the possibility of genetic testing for Familial Adenomatous Polyposis versus other polyposis syndromes. DNA sample was extracted foran Adenomatous Polyposis Coli (APC) gene testing and a comprehensive colon cancer panel testing; however, it was deferred due to financial constraints.

Due to a need for recurrent transfusions, she underwent a prophylactic laparoscopic total procto-colectomy with ileoanal pull-through and ileostomy formation. Total colon was removed along with the appendix, which were then sent for histopathology which revealed a well differentiated adenocarcinoma arising in a background of Familial Adenomatous Polyposis Coli Syndrome (Figure-1). Tumour invaded the muscularis propria with no perineural or vascular involvement. Nine lymph nodes were recovered which were all tumour free. All resection margins were tumour free. Final histopathological staging was pT2 N0 M0. Postoperatively, she was initially kept nil per oral and her diet was then escalated. However, on the 4th post-operative day, she developed post-operative ileus and had to be managed conservatively with a bowel decompression. She improved within 24 hours and was discharged on the 8th post-operative day. She remained well upon follow ups in the clinic till 27th September, 2018.

Discussion

Familial Adenomatous Polyposis (FAP) is one of the most common hereditary syndromes associated with Colorectal Cancer. FAP has an estimated prevalence of 3 in every 10,000 live births, with a relatively equal worldwide distribution among males and females.[1,2] In the paediatric age group, colorectal carcinoma accounts for less than 1 % of all the paediatric malignancies, with an annual incidence of 1 per million.[3]

The severity of colorectal carcinoma manifestations in the FAP is directly associated with the location of the APC gene. Progression to colorectal cancer (CRC) occurs in most patients by their fourth decade of life.[4]

Diagnosis of FAP relies on the presence of greater than 100 polyps within the colon on endoscopic examination.

Alternatively, an attenuated form of the disorder also exists, this is characterized by less than 100 colonic polys, which develops at a later age than the classical FAP (average age 36 years) and it tends to involve the colon more proximally than its classic counterpart. Progression to colorectal carcinoma in classical FAP reaches 100% while progression reaches 80% in the attenuated version of the FAP. Despite the earlier presentation of our patient with FAP, progression to colorectal carcinoma had already ensued. This earlier onset of the disease proved challenging, as the incidence of FAP in those less than 10 years of age is rare.[5] Incidence rates for Pakistan are not known due to resource constraints and non-availability of the genetic testing required.

Our patient may have had the attenuated form of the disorder, since less than 100 polypoid growths were found (Figure-1), however this does not explain the earlier presentation of the patient with progression to colorectal carcinoma.

Treatment strategies for FAP includes two primary surgical options; one being subtotal colectomy with ileorectal anastomosis (IRA) and the other total proctocolectomy with ileal pouch-anal anastomosis (IPAA).[6] Given the dysplastic nature of the polyps in our patient, we decided it best not to leave a rectal remnant behind for fear of the disease progression. Although, the reported incidence for colorectal carcinoma in the rectal remnant in patients with FAP after IRA is only 5.5%, this would still require endoscopic surveillance.[7]

Although, IRA is technically more difficult, we opted to perform the laparoscopy due to the localized nature of the disease. Laparoscopic total colectomy was first defined in 1991[8] and although, its role in adults is well-defined, its inherent role in paediatric patients is still being evaluated.

However, it has become the mainstay of the treatment in well-equipped and high out-put centres for the treatment of refractory ulcerative colitis and FAP. Recent literature reveals similar surgical outcomes between both surgical modalities, in terms of postoperative small bowel obstruction, post-operative abdominal or pelvic abscess, anal stricture requiring dilation, wound infection, other complications or time to complication. There is however, an improved outcome in terms of cosmesis (Figure-2) and lesser incidence of pouchitis.[9,10]

Conclusion

Familial Adenomatous Polyposis Syndrome can present in the first decade of life with an earlier progression to colorectal carcinoma then that reported previously. In such cases, genetic testing is crucial to ascertain the exact type of mutation in the APC gene to guide screening of at-risk family members. As the progression to colorectal carcinoma is 100% prophylactic total proctocolectomy should be advised as soon as possible. Laparoscopic total colectomy has been increasingly being adopted for this purpose with equivocal results achieved when compared to open procedures. Laparoscopic total colectomy performed safely results in the paediatric population with improved cosmesis.

Disclaimer: None to declare.

Conflict of Interest: None to declare.

Funding Sources: None to declare.

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