Colonic Polyposis: Experience in Seven Filipino Adolescents

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Abstract

We report seven adolescents presenting with hematochezia. Five had a family history of colonic polyps or cancer. Colonoscopy showed either multiple pedunculated and/or sessile polyps with partial or total colonic involvement. Polyp histopathology was of the juvenile retention, hamartomatous or tubular adenomatous type. Total proctocolectomy was done in four patients, two underwent repeated polypectomy, and one is awaiting further treatment. An increased awareness of colonic polyposis is important due to the risk of malignant transformation.

Key Words: hematochezia, colorectal carcinoma, prolapsed rectal mass

Introduction

Colonic polyps are grossly visible protrusions from the mucosal surface of the large intestine and commonly present with rectal bleeding, abdominal pain, diarrhea and rectal prolapse. In children, 94% of colonic polyps are isolated juvenile polyps.[1,2] However, some patients may have more than five (multiple) polyps which are mostly located in proximal part of the colon,[2] a condition called colonic polyposis or *polyposis coli*.[3] The true incidence of polyps in childhood remains unknown, and in all studies, a male preponderance is noted.[4,5] All ethnic groups can be affected.[6]

Cases of colonic polyposis are classified histologically as neoplastic (benign or malignant) or non-neoplastic (inflammatory or hamartomatous). There are three main polyposis syndromes that share the features of an autosomal dominant mode of inheritance and an increased risk of colorectal cancer: juvenile polyposis (JP), familial adenomatous polyposis (FAP), and Peutz-Jeghers polyposis (PJP).[7] Histologically, an adenomatous polyp has proliferating epithelial tubules with diminution in the number of goblet cells, crowding of nuclei and increase in the number of mitotic figures—features characteristic of neoplasm. On the other hand, juvenile polyps are hamartomas that develop from an abnormal collection of tissue elements that are otherwise normally present at this site. They have a distinctive cystic architecture, mucus-filled glands, prominent lamina propria and a proliferation of inflammatory cells. These features lead to overlapping terminology, including retention, hyperplastic and inflammatory polyps, depending on the dominant histological findings. Muscle fibers and the proliferative characteristics of adenomas are typically not seen in juvenile polyps.[6,8]

Colonic polyposis in childhood is an uncommon condition with case series reported from India,[4,9] Thailand,[10] Pakistan,[5] Japan,[11] and other countries like Israel,[12] USA,[13] and Finland.[7] At present, there are no reports on the disease in the Philippines. In the last two years, we have seen and diagnosed seven adolescents with colonic polyposis, all with an initial presentation of hematochezia, diarrhea and abdominal pain. The colonic polyps were detected on colonoscopy and confirmed on histopathology. We present their clinical profile and management, along with a review of literature on colonic polyposis.

Consent was obtained from the families of the patients for inclusion in this case series. A summary of all the cases is presented in Table 1.

Cases 1 and 2

Two siblings, females 11 and 13 years of age, consulted for severe abdominal pain, hematochezia and prolapsed rectal mass. Their father had similar symptoms and died of gastric cancer at 34 years of age.

The younger sibling was noted to have a prolapsed rectal mass at 2 years old. At 4 years, she had hematochezia. At that age, proctosigmoidoscopy was done to remove a large polyp which was found to be a juvenile retention polyp by histopathology. Upper endoscopy revealed multiple gastric and duodenal polyps. Hematochezia recurred at 8 years of age and again at 11 years, hence the most recent consult. On physical examination, the patient was pale, with moderate malnutrition (BMI Z=-2). She had multiple skin pigmentations most prominent on the lips and buccal mucosa (Figures 1A to 1C) and a palpable rectal mass. At the time of this writing, the plan was to do a total colonoscopy and possible polypectomy.

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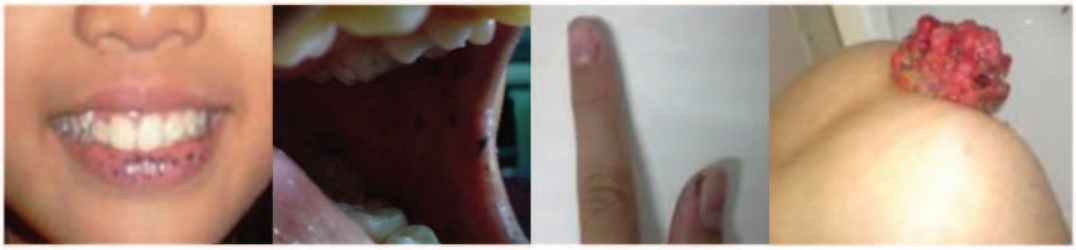
Table 1. Summary of individual cases

	CASE 1	CASE 2	CASE 3	CASE 4	CASE 5	CASE 6	CASE 7
Age	11	13	15	16	16	14	17
Sex	F	F	F	M	F	M	M
Duration of symptoms	9 years	8 years	1 year	3 years	2 weeks	9 years	4months
Associated illness	Skin pigmentations	Skin pigmentations Abnormal uterine bleeding	none	none	none	Hepatoblastoma	Skin pigmentations
Family history of colon polyps or cancer	yes	yes	none	yes	yes	yes	no
Colonoscopy findings	Proctosigmoidoscopy: large rectosigmoid polyp	Intra-op: Multiple small and large bowel polyps	Multiple pedunculated polyps of the whole colon	Multiple sessile and pedunculated polyps of the whole colon	Multiple pedunculated polyps from rectum to transverse colon	Multiple sessile polyps of the whole colon	Multiple sessile and pedunculated polyps whole colon
Histopathologic findings of the colonic polyps	Juvenile retention polyp	Hamartomatous polyps	Juvenile retention polyps	Juvenile retention polyps	Tubular adenomatous polyps	Tubular adenomatous polyps and inflammatory pseudopolyps	Hamartomatous polyps
Surgical intervention	awaiting	Explore lap reduction of intussusception	Total proctocolectomy with ileoanal anastomosis	Total proctocolectomy with ileoanal anastomosis	Total proctocolectomy with ileoanal anastomosis	Total proctocolectomy with ileoanal anastomosis	none
Hemoglobin (nv:120-180 g/L)	83	69	82	78	75	132	102
Albumin (nv: 34-50 g/L)		31	15	29	15	38	
Potassium (nv: 3.6-5.2 mmol/L)		3.4	2.7	3.8	2.4	4.5	
Radiologic studies	Barium enema: polypoid lesion sigmoid colon	Transrectal UTZ: endometrial mass, consider endometrial polyp	Barium enema with SIS: rectal masses	UGIS with SIS: normal	Barium enema: non specific colitis vs polyposis	UGIS with SIS: unremarkable	Abdominal CT SCAN: non-enhancing filling defect with well-define borders alongside the lesser curvature of the stomach
Esophagogastro duodenoscopy (EGD)	Gastric and duodenal polyps	Gastric polyps	Normal	Normal	Normal	Gastric polyp, nodular / scalloping of the duodenal mucosa	Multiple sessile and pedunculated gastric polyps

The elder sibling presented with a prolapsed rectal mass at 5 years of age. However, no consult was done until she was 11 years old, after she developed severe abdominal pain, hematochezia and increased uterine bleeding. She subsequently underwent surgery for manual extraction of jejunojejunal and colocolic intussusceptions. Histopathologic examination revealed hamartomatous jejunal and colonic polyps. Transrectal ultrasound also showed an endometrial polyp. Recurrence of hematochezia, prolapsed rectal mass and pallor prompted the most recent consult. On physical examination, the patient was pale, with moderate wasting (BMI Z=–2). She had multiple skin pigmentations prominent on the lips and buccal mucosa and a palpable fleshy rectal mass (Figure 1D). Upper and lower endoscopy revealed multiple sessile and pedunculated gastric and colonic polyps. Polypectomy was done on polyps greater than 1 cm



A B C D

Figure 1. Physical examination findings of patients with colonic polyposis. Mucocutaneous pigmentation of lips (A) buccal mucosa (B) and fingers(C), and prolapsed rectal mass (D).

in size; these were found to be of the hamartomatous type. At the time of this writing, the plan for this patient was serial colonoscopy and polypectomy.

Case 3

A 16-year-old male was referred for a three-year history of recurrent hematochezia associated with diarrhea and prolapsed rectal mass. Two of his siblings had similar symptoms and died at the age of 14 years with no known diagnosis. A maternal grandmother and maternal aunt had colonic carcinoma. On physical examination, the patient was pale but not wasted (BMI Z = 0 to 2). The patient was found to have hypoalbuminemia. Colonoscopic examination revealed multiple polyps from rectum up to cecum, described as sessile and pedunculated, small to large in size. Most of the polyps were dark purple, though some were pale. The polyps ranged from 0.5 mm to 1.5 cm in size with stalks up to 8 cm (Figures 2A and 2B), and were arranged in groups. The polyps were found to be juvenile retention polyps.

The patient underwent total proctocolectomy and ileoanal anastomosis but subsequently required an ileostomy due to an intraabdominal abscess (Figure 2C). He eventually improved and, at the time of this writing, was able to tolerate a regular diet.

Case 4

A 15-year-old female was referred to our institution due to massive hematochezia and pallor. She had a one-year history of diarrhea, abdominal pain and hematochezia with a reducible rectal "mass". Previous proctosigmoidoscopy and polypectomy done in another hospital revealed juvenile polyps. There was no family history of colonic polyps. On physical examination, the patient was pale and emaciated (BMI Z = <-3), and had bipedal edema. Laboratory findings revealed anemia, hypoalbuminemia and hypokalemia. Colonoscopic examination revealed multiple pedunculated polyps, with some ulcers, in clumps from rectum to cecum. The polyps were diagnosed as juvenile retention polyps (Figure 3A).

The patient underwent total proctocolectomy with ileoanal anastomosis (Figure 2D). Postoperatively, she continued to have hypoproteinemia, hypoalbuminemia and diarrhea and eventually died secondary to (E. coli) septicaemia.

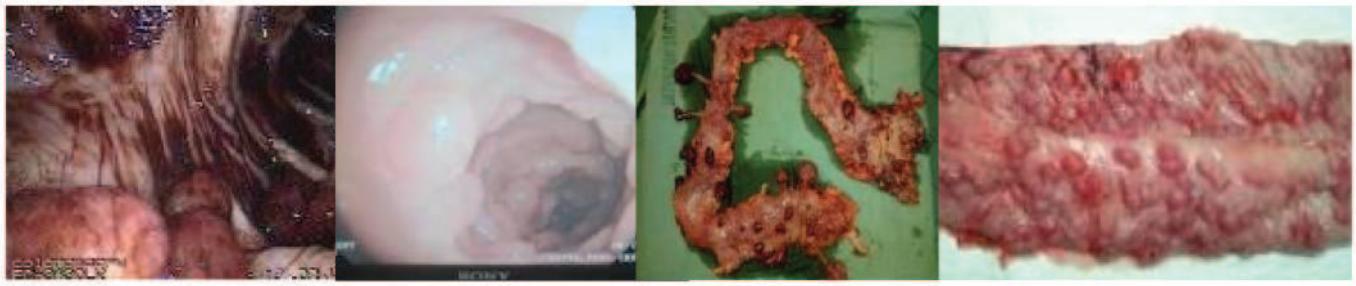
Case 5

A 16-year-old female consulted with a two week history of hematochezia associated with abdominal pain, vomiting and a reducible rectal mass (Figure 1D). She was initially managed as a case of intestinal amebiasis but persistence of symptoms prompted consult at our institution. Her maternal grandfather had colonic cancer. On admission, patient was pale with bipedal edema and was severely malnourished (BMI Z = < –3).

Laboratory examinations revealed anemia, hypoalbuminemia and hypokalemia. Colonoscopy revealed multiple sessile to pedunculated polyps from rectum to mid transverse colon sparing the ascending colon. She was diagnosed as having tubular adenomatous polyps (Figure 3B). She underwent total proctocolectomy with ileoanal anastomosis and was given elemental formula and total parenteral nutrition prior to and after surgery. Postoperative complications included nosocomial pneumonia and wound dehiscence, with persistent hypoalbuminemia, hence a prolonged hospital stay and recovery time.

Case 6

A 14-year-old male consulted with a nine-year history of blood-streaked loose stools and a two-year history of these symptoms associated with abdominal pain. He was diagnosed with hepatoblastoma at two months old and was treated with chemotherapy and resection of the hepatic mass. A maternal great grandfather had colonic cancer and a maternal uncle had a history of diarrhea and bloody stools, and underwent colectomy, although the diagnosis was undisclosed. On admission, physical examination (BMI Z = 0 to 2) and laboratory investigations were unremarkable. Colonoscopy revealed multiple sessile polyps from rectum to cecum. These polyps were of the tubular adenomatous



A B C D

Figure 2. Colonoscopic and gross specimen of patient with colonic polyps. Colonoscopy showing multiple pedunculated (A) and sessile polyps (B) ranging in size from 0.5 to 1.5 cm and stalk of up to 8 cm. Resected gross specimen during total colectomy(C & D)

type. Hyperplastic antral polyps were also seen on esophagogastroduodenoscopy.

A total proctocolectomy and ileoanal anastomosis was done. The postoperative course was uneventful.

Case 7

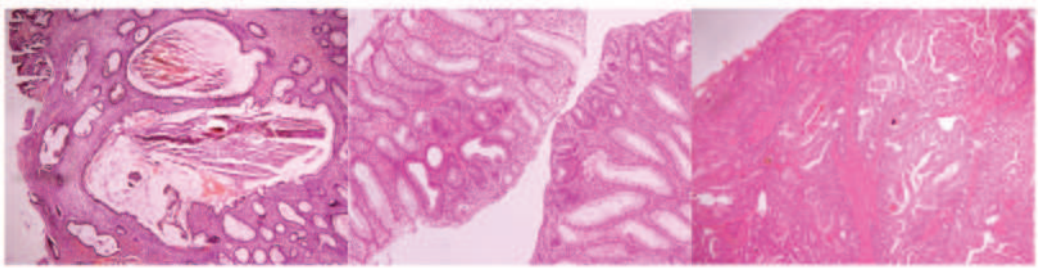
A 17-year-old male was referred for a four month history of hematochezia with occasional episodes of abdominal pain. There was no weight loss nor any changes in bowel habits. The patient was initially diagnosed as having peptic ulcer disease but did not experience relief by antacids. There was no family history of a similar illness or colon cancer. On physical examination, there were multiple skin pigmentations prominent on the lips, buccal mucosa and fingers, and a palpable fleshy rectal mass. Abdominal CT scan demonstrated a non-enhancing filling defect with well-defined borders along the lesser curvature of the stomach. Upper and lower endoscopy revealed multiple gastric and colonic polyps of the tubular adenomatous and hamartomatous type (Figure 3C). At the time of this writing, the patient was asymptomatic and had been advised serial endoscopy and polypectomy.

Discussion

We report for the first time a case series of Filipino adolescents who initially presented with hematochezia and were eventually diagnosed to have colonic polyposis after colonoscopy and histopathologic examination of the polyps. Three of these patients (cases 1, 2 and 7) were diagnosed to have Peutz–Jegher Syndrome (PJS) in the presence of gastrointestinal polyps and mucocutaneous pigmentations, the latter present in 95% of patients with this condition.[14]

Similar to previous reports, our patients initially presented with hematochezia, weight loss, diarrhea and abdominal pain. Of the seven, five had pallor and a prolapsed rectal mass and four had concomitant malnutrition and bipedal edema. The latter signs were brought about by protein-losing enteropathy from the interference of the presence of the polyps on the intestinal absorptive capacity.[9] This constellation of findings should alert the physician on the need for further work-up in these patients, including a full-length colonoscopy.[15] Often, in our setting, these patients are treated as a case of dysentery or amebiasis.

Of interest is that five of our patients had a family history of colonic polyps or cancer and one was previously diagnosed to have hepatoblastoma. Family studies have demonstrated genetic mutations, including SMAD4 or BMPRIA, in 40 to 60% of patients with juvenile polyposis.[1] It has been reported that only 75% of patients diagnosed with juvenile polyposis will have a family history and the rest will have *de novo* mutations.[16,17] This is demonstrated in the



A B C

Figure 3. Histopathologic findings of patients with colonic polyposis. Juvenile retention polyps (A); adenomatous polyps (B) and hamartomatous polyps (C)

third and seventh cases, who had no family history of polyposis. On the other hand, there is an established link between the adenomatous polyposis coli (APC) gene at the long arm of chromosome 5 and the tubular adenomatous polyps noted in our cases 3 and 4.[1,18] The APC gene has also been associated with extracolonic tumors including hepatoblastoma and gastric polyps, which one of our patients had.[8,19] Moreover, *LKB1 (STK11)* is the only gene known to be associated with autosomal dominant Peutz–Jeghers Syndrome; [8,14,20] this can explain the familial occurrence seen in cases 1 and 2, in which the father was also afflicted.

At the time of this writing, four of our patients had undergone total proctocolectomy with ileoanal anastomosis; two were undergoing repeated polypectomy and another was waiting for a total colonoscopy. The main indication for a proctocolectomy is that the size and number of polyps (>20) are difficult to control with repeated colonoscopy and polypectomy.[1,21] It is also indicated in the presence of intractable signs and symptoms including anemia, hypokalemia and hypoalbuminemia. While virtually all patients with familial adenomatous polyps will develop colorectal carcinoma if left untreated, hence the need for prophylactic colectomy, the data is insufficient to justify a similar procedure in those with juvenile polyposis solely because of the risk of carcinoma.[1,4,12,16] With respect to PJS, the immediate risk to the patient is the development of small bowel intussusceptions and obstruction as experienced in case 2. This could have been prevented with surveillance, considering that there is a family history.[14] The treatment recommendation for PJS with symptomatic polyps is elective laparotomy with intraoperative enteroscopy. The same holds true for asymptomatic polyps which are pedunculated and greater than 1 cm in size.[14] Patients with PJS also have a 30% and 50% lifetime risk of developing colon and breast cancer, respectively.[8]

The first degree relatives of our patients were advised regarding early screening and treatment. It has been suggested that family members of patients with juvenile or adenomatous colonic polyposis should be screened by the age of 15 years[4,13] and those with PJS at eight years old.[14] If positive, polypectomy should be attempted and the patient should return yearly for upper and lower endoscopy until free of polyps. If results are negative, upper and lower endoscopy is to be repeated every three years until the age of 40.[1,4] The rationale for this is that the average age of occurence for colon cancer is at 44 years.[13]

In summary, this case series showed an increased frequency of colonic polyposis in the Philippines. This may mean a true increase or an improved awareness of the disease. Colonic polyposis should be included in the differential diagnosis of any child with unexplained rectal bleeding due to the risk of malignant malformation.

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References

1. Durno CA. Colonic polyps in children and adolescents. Can J Gastroenterol. 2007;21(4):233–9.
2. Haghi Ashtiani MT, Monajemzadeh M, Motamed F, et al. Colorectal polyps: a clinical, endoscopic and pathologic study in Iranian children. Med Princ Pract. 2009;18(1):53–6.
3. Veale AM, McColl I, Bussey HJ, et al. Juvenile polyposis coli. J Med Genet. 1966;3(1):5–16.
4. Upadhyaya VD, Gangopadhyaya AN, Sharma SP, et al. Juvenile polyposis syndrome. J Indian Assoc Pediatr Surg. 2008;13(4):128–31.
5. Mandhan P. Juvenile colorectal polyps in children: experience in Pakistan. Pediatr Surg Int. 2004;20(5):339–42. Epub 2004 May 18.(Abstract)
6. Mougenot J, Olschwang S, Peuchmaur M. Intestinal Polyps and polyposis. In: Kleinman R, Sanderson I, Goulet O, Sherman, P, Mieli-Vergani G, Shneider B, eds. Walker's Pediatric Gastrointestinal Disease, 5th ed. 2008. Hamilton: BC Decker, Inc. pp. 637–650.
7. Jarvinen HJ. Genetic testing for polyposis: practical and ethical aspects. Gut. 2003;52 (Suppl II): ii19–ii22.
8. Larsen Haidle J, Howe JR. Juvenile Polyposis Syndrome.In: Pagon RA, Bird TD, Dolan CR, Stephens K, editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2003 May 13 [updated 2008 Sep 09].
9. Poddar U, Thapa BR, Vaiphei K, et al. Juvenile polyposis in a tropical country. Arch Dis Child. 1998;78(3):264–6.
10. Ukarapol N, Singhavejakul J, Lertprasertsuk N, et al. Juvenile polyp in Thai children--clinical and colonoscopic presentation. World J Surg. 2007;31(2):395–8.(Abstract)
11. Yoshida H, Tomichi N, Mori S, et al. Juvenile polyposis coli. A case report. Jpn J Surg. 1982;12(4):274–9.
12. Rozen P, Baratz M. Familial juvenile colonic polyposis with associated colon cancer. Cancer. 1982;49(7):1500–3.
13. Brosens LA, van Hattem A, Hylind LM, et al. Risk of colorectal cancer in juvenile polyposis. Gut. 2007;56(7):965–7.
14. Hinds R., et al. Complications of childhood Peutz–Jeghers Syndrome:implications for pediatric screening. J Pediatr Gastroenterol Nutr. 2004;39(2):219–20.
15. Williams CB, Laage NJ, Campbell CA, et al. Total colonoscopy in children. Arch Dis Child. 1982;57(1):49–53.
16. Adolph VR, Bernabe K. Polyps in children. Clin Colon Rectal Surg. 2008;21(4):280–5.
17. Erdman SH. Pediatric adenomatous polyposis syndromes: an update. Curr Gastroenterol Rep. 2007;9(3):237–44.
18. Beech D, Pontius A, Muni N, et al. Familial adenomatous polyposis: a case report and review of the literature. J Natl Med Assoc. 2001;93(6):208–13.
19. Roth S. Molecular genetic background of juvenile polyposis. Academic Dissertation. ISBN 952-91-1792-2 Cosmoprint OY, Helsinki 2000.
20. Giardiello FM, Petersen GM, Brensinger JD, et al. Hepatoblastoma and APC gene mutation in familial adenomatous polyposis. Gut. 1996; 39(6):867–9.
21. Campbell WJ, Irwin ST, Parks TG. The surgical management of familial adenomatous polyposis in Northern Ireland. Ulster Med J. 1991;60(2):193–8.