ORIGINAL RESEARCH PAPER

Surgery

KEY WORDS: Juvenile polyposis coli, juvenile polyposis syndrome, hamartomatous polyp, total proctocolectomy and ileal-pouch anal anastomosis, j-pouch

A CASE ON JUVENILE POLYPOSIS COLI

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ABSTRACT

INTRODUCTION Juvenile polyposis syndrome is an inherited autosomal dominant syndrome characterized by hamartomatous intestinal polyp. Symptoms include acute or chronic gastrointestinal bleed, abdominal pain, diarrhea, iron deficiency anemia, prolapsed rectal polyp, intestinal obstruction and intussusception. Diagnosis is by colonoscopy. Surgery involves polypectomy and colectomy. **AIMS** To present a case of patient having juvenile polyposis coli. **MATERIAL AND METHOD** A 4 year old male patient having juvenile polyposis coli proven using colonoscopy biopsy showed multiple polyp in colon and no extra colonic polyp with no malignancy. Patient was planned for total proctocolectomy and ileal-pouch anal anastomosis. **RESULT** Juvenile polyposis syndrome is an inherited disorder so it is not possible to prevent. Treatment is polypectomy or colectomy. **DISCUSSION** Juvenile polyposis syndrome occur due to two genes, SMAD4 and bone morphogenetic protein receptor type 1A- BMPR1A. Individuals who meet clinical criteria of juvenile polyposis syndrome should have genetic testing. It confirms juvenile polyposis syndrome and serves to counsel at risk family members. Approx. 20 to 50% patients have no family history and occur de novo.

INTRODUCTION

Juvenile polyposis syndrome is an inherited autosomal dominant syndrome characterized by hamartomatous intestinal polyp. It is clinically diagnosed when

- there are five or more juvenile polyps in colorectum,
- multiple juvenile polyps throughout gastrointestinal tract,
- Any number of juvenile polyps with family history.

Symptoms include acute or chronic gastrointestinal bleed, abdominal pain, diarrhea, iron deficiency anemia, prolapsed rectal polyp, intestinal obstruction and intussusception.

Diagnosis is by colonoscopy. Screening should begin between 12 to 15 years. Average age of diagnosis is 34 years. If there are no polyp repeat colonoscopy every 2 to 3 years. When polyp are present and removed, colonoscopy should be done annually.

Surgical indication:

- High grade dysplasia or cancer
- Polyp burden cannot be effectively managed endoscopically
- Persistent blood loss resulting in anemia and hypoalbuminemia
- Prophylactic colectomy: family history of colorectal carcinoma or poor surveillance compliance.

Surgery:

1. Subtotal colectomy with ileo-rectal anastomosis
2. Segmental colectomy
3. Total proctocolectomy with ileo-analpouch anastomosis.

AIMS AND OBJECTIVE

To present a case of 4 year old male having juvenile polyposis coli without any significant family history.

CASE

A 4 year old male came to civil hospital Ahmedabad with complaint of persistent diarrhea and bleeding PR since 2 years. Patient was a diagnosed case of juvenile polyposis syndrome.

Colonoscopy done on 12th September 2020, in private clinic at Jhansi showed Rectum, sigmoid and descending colon: varying size pedunculated and sessile polyp with ulcerations Transverse and ascending colon: polyps which are less severe with normal mucosa.

Polypectomy was done of one large rectal polyp.

Colonoscopy done on 16th October 2020 in private clinic in Ahmedabad showed multiple polyps of 6mm to 1.4cm throughout colon. Left colonic polyp multiple with some having superficial ulceration. Two largest polyp removed with cautery and snare. Right colon showed two small polyp.

Both outside colonoscopy were diagnostic of juvenile polyposis coli.

On presentation to civil hospital Ahmedabad in April 2022, patient had diarrhea and bleeding pr. No symptoms of abdominal pain, vomiting, hematemesis. Patient was advised contrast enhanced CT scan abdomen plus pelvis and repeat colonoscopy. Cect was suggestive of multiple soft tissue density lesions seen arising from bowel wall projecting into lumen in ascending, transverse, descending, sigmoid colon and rectum suggesting multiple polyps. Colonoscopy on 19th April, 2022 showed multiple polyp from rectum to splenic flexure (Inadequate bowel preparation). Biopsy taken showed fragments of normal mucosa with polypoid ulceration and inflammation. Polyp lined by colonic epithelium with hyperplastic crypts suggestive of benign inflammatory polyp. Sonography also showed multiple pedunculated polyp with internal vascularity in ascending, transverse, descending, sigmoid colon and rectum.

Patient was admitted to civil hospital Ahmedabad on 3rd may 2022 with same symptom.

Systemic examination:
Temperature normal
Pulse 110/min
Bp 108/70 mmHg
Spo2 98 on room air
RS BLAE+

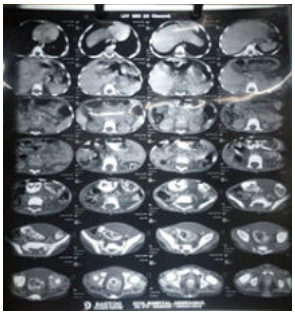
CVS S1S2+
CNS conscious
P/A soft non tender

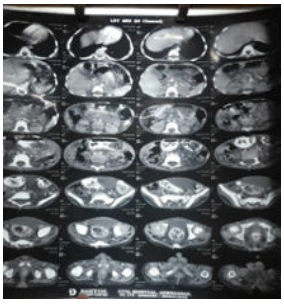
Colonoscopy was done on 4th may, 2022 which showed

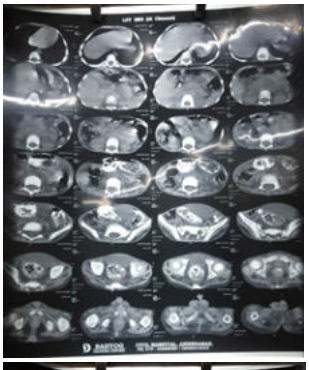
Anal canal, rectum and sigmoid: multiple varying size sessile polyp

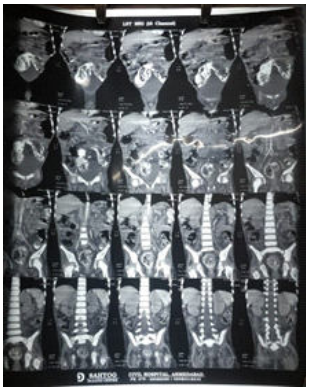
Descending colon: multiple large sized polyp
Transverse and ascending colon: multiple large sized polyp. Transverse colon biopsy taken was suggestive of hyperplastic polyp with chronic inflammatory cells in lamina propria with no evidence of malignancy or dysplasia.

Upper gastrointestinal endoscopy done on 12th may, 2022 showed duodenitis in its 1st part with no evidence of extra-colonic polyps. Biopsy taken showed duodenal mucosa chronic inflammation with no polyp/dysplasia/malignancy.









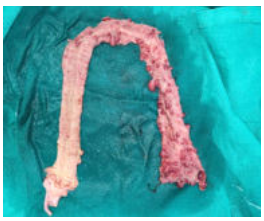
MANAGEMENT

We planned total proctocolectomy with ileal pouch anal anastomosis with diverting loop ileostomy by creating a J pouch. In lithotomy position, anal mucosa cut 1 cm above dentate line. Rectum mobilized. Midline laparotomy incision placed and total proctocolectomy done after mesentery ligation. Terminal ileum is made into a J pouch of approx. 8 cm and sutured using vicryl 3-0. Terminal end of pouch is sutured with anal mucosa with silk 4-0. Diverting loop ileostomy done. Drain was placed in pelvis. No intra op complication present. Post op patient developed hypotension for which inotropes were started. ECG showed sinus rhythm with RBBB. Post-operative 2D Echo was normal.

He was gradually weaned off inotropes. Drain was removed on POD 12

Patient came for follow up with no complaints and healthy stoma.

Biopsy obtained from part of ileum, ileo-caecal junction, appendix, colon specimen showed multiple polyp from colon and rectum with histology of juvenile polyposis coli with cystic and markedly dilated glands, some of which contained inspissated mucin and edematous inflamed lamina propria with focal mild dysplasia without any malignancy with chronic appendicitis and resected 11 lymph nodes showed sinus histiocytosis with no tumor cell.



CONCLUSION

This case was of 4year old male having persistent bleeding PR and diarrhea and colonoscopy showed multiple polyps with history of polypectomy. Ileal pouch anal anastomosis was planned. Juvenile polyposis syndrome is an inherited disorder so it is not possible to prevent. Ileostomy closure is to be planned on a later date.

DISCUSSION

Etiology: Juvenile polyposis syndrome occur due to two genes, SMAD4 and bone morphogenetic protein receptor type 1A- BMPR1A which occur in 50 to 60% of patients where they affect BMP-TGF beta signaling pathway. Patients with SMAD4 have higher risk of gastric polyp and gastric cancer than BMPR1A mutation. It belongs to group of hamartomatous polyps, Peutz-Jeghers syndrome and Cowden syndrome.

Clinical criteria and Incidence: diagnosis confirmed if any 1 of following is present:

- there are five or more juvenile polyps in colorectum,
- multiple juvenile polyps throughout gastrointestinal tract,
- Any number of juvenile polyps with family history.

Individuals who meet clinical criteria of juvenile polyposis syndrome should have genetic testing. It confirms juvenile polyposis syndrome and serves to counsel at risk family members. Approx. 20 to 50% patients have no family history and occur de novo. Polyps usually begin in first decade of life and occur predominantly in colorectum (98%), stomach (14%), duodenum (7%), jejunum and ileum (7%). Patient with juvenile polyposis syndrome has risk of 10-38% of developing Colorectal and gastric cancer.

Diagnosis: Colonoscopy guided biopsy. Biopsy shows cystically dilated glands filled with mucuc lined by cuboidal or columnar epithelium, abundance of edematous lamina

propria with inflammatory cells, and absence of smooth muscle in the stroma.

DIFFERENTIAL DIAGNOSIS

Familial Adenomatosis Polyposis- patients with jps may have concomitant adenomatous polyps which can cause misdiagnosis as fap. Pathologically, its difficult to differentiate between juvenile polyp with dysplasia and adenoma. Fap has small sessile polyp whereas jps has pedunculated polyps. Presence of pedunculated polyp and pathological examination of more polyp will help in accurate diagnosis.

Surgery:

1. Subtotal colectomy with ileo-rectal anastomosis
2. Segmental colectomy
3. Total proctocolectomy with ileo-analpouch anastomosis.

Continued surveillance post-surgery is required since polyps may develop in ileum or rectum.

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