JUVENILE POLYPOSIS SYNDROME IN A YOUNG PATIENT: A RARE CASE REPORT

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ABSTRACT

Introduction

Juvenile polyposis syndrome prevalence is 1 in 16,000 to 1 in 100,000 which usually present at the age of 20 years. The tumor is likely to change into malignant condition in 20% of cases. Here we present this rare syndrome in a 16-year boy.

KEYWORDS

Hamartomatous polyps, juvenile polyposis syndrome, malignant.

INTRODUCTION

Juvenile polyposis syndrome is a rare autosomal dominant disease characterized by different size of polyps, most commonly seen in the colorectal segment of large intestine.[1] The term "juvenile" indicates for the type of polyp not the age of the patient when the polyp develops hence it can be seen in any age group.[2] More than 20% of Juvenile polyposis syndrome patients are found to have inborn defect of different organs like Meckel's diverticula with umbilical fistula, undescended testes, unilateral renal agenesis, split uterus, arterionevoushaemangiomas, stenosis of the pulmonary valve, macrocephaly and intermittent porphyria.[3]

In 1975 the diagnostic criteria for juvenile polyposis syndrome was established. This criteria was revised by Jass et.al.[4] in 1988 mentioning one of following three criteria must be present:

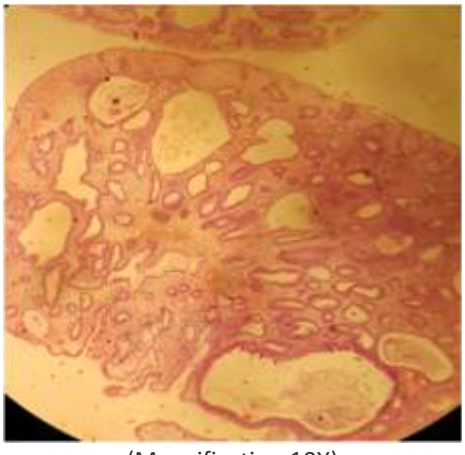
1. >5 juvenile polyps in the large intestine or/and
2. Multiple juvenile polyps throughout the gastrointestinal tract or/and
3. Any number of juvenile polyps with a family history of juvenile polyposis

Positive family history has been associated with 20 – 50% of affected patients.[3] Children with Juvenile polyposis are susceptible to gastrointestinal cancers.[5]

The common presentations are anemia most commonly due to chronic recurrent gastrointestinal bleeding, and diarrhea however some cases had been associated with surgical abdomen like rectal prolapse and intussusception, or medical complications like protein losing enteropathy, starvation and malnutrition.[6]

CASE REPORT

A 16 year old boy presented with anemia, hypoalbumenia and bleeding per rectum associated with mass coming out per anus for 2 to 3 years. He had alternating diarrhea and constipation with episodic mild abdominal pain. Colonoscopy showed multiple polyps of varying size of both sessile and pedunculated in the rectum and entire colon. Polyps from rectum were sent for histopathological examination revealing benign adenoma. Clinical diagnosis of Familial Adenomatous Polyposis was made and counseled for pan-colectomy.



(Magnification 10X)

Figure 1: Low power view reveals polypoidal tissue having cystic like glands with budding and branching.

Macroscopy examination on cut opening of specimen multiple colorectal polyps of varying sizes on the mucosal surface was present. The outer surface of polyp was glistening. It was smooth to touch. Cut surface revealed solid gray white to tan red with few cystic spaces contains jelly like material of size ranging from 0.1 to 0.2cm.

Microscopy examination of the representative section from entire tissue showed multiple polypoidal tissue revealing tortous gland with budding and branching. (Figure 1) The glands were lined by mucus secreting columnar cells and contain eosinophilic material. The stroma between the glands contained acute and chronic inflammatory cells as well as granulation tissue and hemorrhage. The overlying lining epithelium was partly eroded and erosion free areas were lined by tall columnar cells. (Figure 2 and 3) Diagnosis of Juvenile Polyposis Syndrome was made on the basis of histopathology.

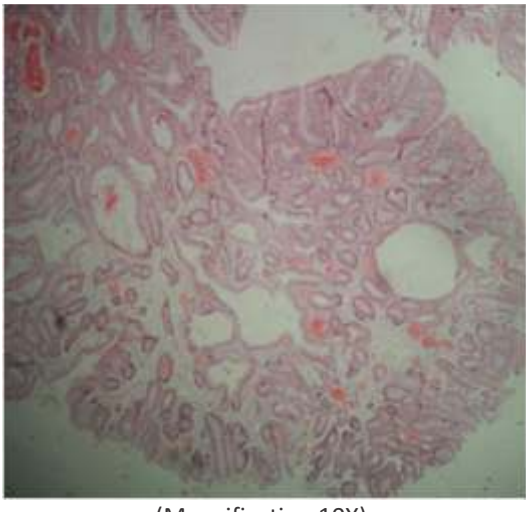


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Figure 2: Low power view reveals polypoidal tissue having cystic like glands with budding and branching.

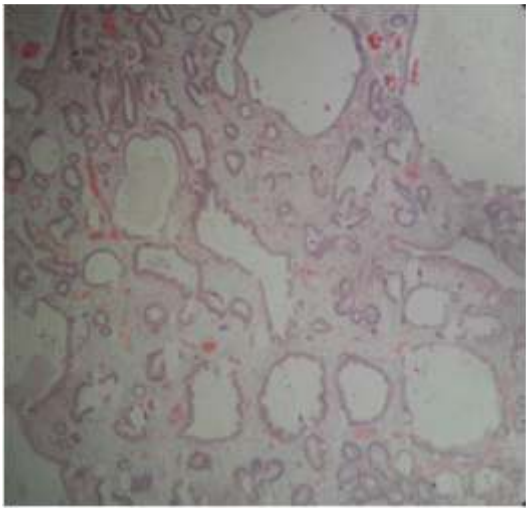

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Figure 3: High power view reveals glands lined by mucus secreting columnar cells. Stroma is edematous and inflammatory cells infiltrates with granulation tissue.

DISCUSSION

Juvenile polyposis syndrome is an autosomal dominant condition diagnosed on occurrence hamartomatous gastrointestinal polyps . Patients with juvenile polyposis syndrome are likely to carry gene carriers to various types of tumors. The hamartomatous polyps may turn into malignant lesions in approximately 20% of cases.[7] Juvenile polyposis syndrome demonstrated different heterozygous mutations most commonly SMAD4 and BMPR1 on chromososme 10q21 located on chromosome 18q21 respectively.[8] Evidence of mutation of SMAD4 gene in the family with one member especially siblings or children affected has theoretical risk of being carriers of 50% [9]. Molecular diagnostics is to optimize prevention, early diagnosis and management at the family level as the chances of malignant transformation is very high.

In our patient there was a family history of rectal polyp and had history of malignancy to cousin. In case of positive family history like the case routine screening colonoscopy is recommended from the age of 10 -12 years, every 2 years until age 40 or beyond if we had screened for genetic mutation. As the mutation detection was out of scope in our settings we strongly recommend colonoscopy screening among all relatives at high risk.[9]

The occurrence of juvenile polyposis syndrome is between 1 in 16,000 and 1 in 100,000.[10] Majority of patient develop symptoms by the time of 20 years old. The polyps for unknown reason are most often grow in the large intestine (colon) and rectum as in our case however rare growth in the stomach and the small intestine has been documented.[10]

Macroscopically it varies in size from 5mm to 50mm and has spherical, lobulated and pedunculated appearance with surface erosion. Microscopically a juvenile polyposis is characterized bypresence of abundant of inflammatory cells at edematous laminapropria with and cyst like glands lined by cuboidal to columnar epithelium that has reactive changes within the dilated gland.[11]

These polyps may be the seat of focus of dysplasia and in some cases; true adenomas were described in their vicinity.[11]Our patient macroscopically and microscopically showed as solitary juvenile polyposis syndrome, neither a contingent of adenomas or dysplasia.

The treatment depends on the clinical presentation, location and number of polyps. Routine colonoscopy screening with endoscopic polypectomyif needed is the most effective treatment of solitary polyps. However surgical intervention like subtotal or total gastrectomy or pan colectomy is recommended in case of multiple number of polyps that minimize the symptoms and to prevent malignancy transformation in future.[12]

Pan colectomy in our patient was definitive cure for rectal bleeding and prevention of development of malignancy.

CONCLUSION

Juvenile polyposis syndrome is a rare disease usually seen among teens but can be fatal or incurable that can be prevented by surgical intervention. The screening of subclinical disease in family with colonoscopy is beneficial.

CONFLICT OF INTEREST

No conflict of interest

FINANCIAL DISCLOSURE

None

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