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Case Report

Juvenile polyposis syndrome with extraintestinal anomalies: report of a rare case with review of literature

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ABSTRACT

Juvenile polyposis coli is a rare condition in children with neoplastic potential having an incidence of about 1 in 1,00,000 population. A minority of such patients have extraintestinal abnormalities like cardiac and pulmonary arteriovenous malformations. Juvenile polyposis is a disorder of hamartomatous polyposis syndrome having a malignant potential. The progression of hamartomatous polyp to carcinoma is still elucidated when compared to the understanding of transformation of an adenomatous polyp into a carcinoma via a gatekeeper defect. Here is the report of a rare case of Juvenile polyposis in a 7 year old boy who presented with bleeding per rectum and prolapsed rectum showing multiple polyps. Patient had undergone surgery for closure of ventricular septal defect and pulmonary valvotomy 3 years back. Proctocolectomy was done and the resected colon showed 40 polyps. Histologically polyps contained benign glandular tissue and one of the larger polyps showed low grade epithelial dysplasia. In this case, there was no positive family history and extraintestinal congenital defects are said to be more common in such sporadic cases.

Keywords: Extraintestinal anomalies, Hamartomatous polyps, Juvenile polyposis coli

INTRODUCTION

Juvenile polyposis syndrome (JPS) belongs to the rare, interesting group of hamartomatous polyposis syndromes with an incidence of 1 in 1,00,000 - 160000. JPS can be familial with an autosomal dominant inheritance or sporadic. The polyps are usually benign with a malignant potential.[1] Sachatello classified them into three subtypes: 1) JPS with polyps localized to the colon; 2) JPS of infancy, this severe form seen in infants and children presents with hypoproteinemia, malnutrition, bloody diarrhoea; 3) larger polyps cause rectal prolapse and intussusception; Generalized JPS, with evenly distributed polyps throughout the gastrointestinal tract.[2] Majority of patients with JPS have a de novo mutation and 11- 20% suffer from other congenital extraintestinal abnormalities which are more common in the sporadic cases.[1,3] We report a case of sporadic JPS in a child with multiple colonic polyps who also had ventricular septal defect and pulmonary stenosis as associated congenital anomalies.

CASE REPORT

A 7 year old boy presented with rectal prolapse and bleeding of five months duration. The prolapsed portion was increasing in size. There was no family history and the patient had undergone surgery for ventricular septal defect and pulmonary valvotomy 3 years back in another hospital. On clinical examination patient had multiple polyps over the prolapsed rectum, microcytic anaemia and blood in stools. Endoscopic examination aided in making the diagnosis of JPS and total proctocolectomy with ileo-anal anastomosis was done. On follow-up the patient is doing well 2 years post-surgery.

Grossly there were two segments of large intestine, longer one measured 64cm and shorter one 10 cm in length along with appendix. The mucosa showed 40 polyps, sessile and pedunculated, largest measuring 3.5 X 3.0 X 0.5cm and smallest 0.5 X 0.5 cm. Surface of the polyps was smooth to lobulated and cut surface showed cystic and hemorrhagic areas (Figure 1). 25 small lymph nodes were dissected.

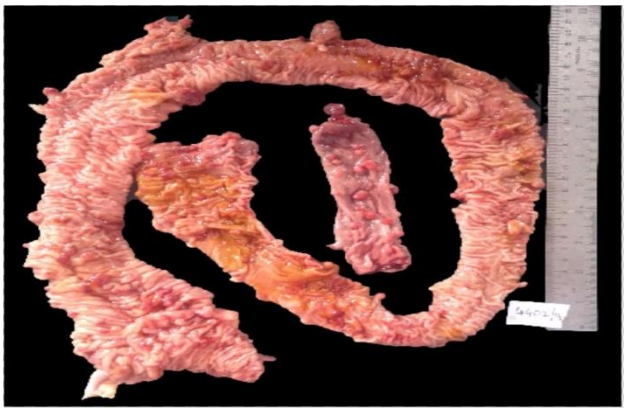

Figure 1: Specimen of large intestine showing multiple sessile & pedunculated polyps having smooth glistening to lobulated surface.

Histopathologically, polyps were covered by tall columnar mucinous epithelium with variable sized mucus filled glands beneath. Some glands were dilated and the stroma showed inflammatory cells and congested blood vessels. Most of the polyps had surface ulceration with granulation tissue.

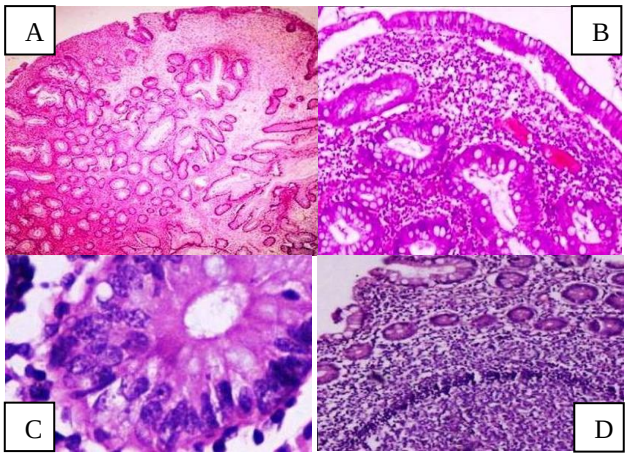

Figure 2: (a) Microscopically polyps showed columnar epithelial lining with variable sized glands. (Haematoxilin & Eosin 40 ×) (b) Inflammatory cells in the stroma (H & E 100 ×) (c) Glandular lining cells exhibiting dysplasia in a larger polyp.(H & E. 400×) (d) Marked lymphoid hyperplasia in the colonic mucosa adjacent to the polyps.(H & E 100 ×).

One of the larger polyp showed adenomatous change and low grade epithelial dysplasia with glandular cells exhibiting moderate pleomorphism, vesicular nuclei, prominent nucleoli and increased mitotic activity. The colonic mucosa adjacent to the polyps and the appendicular wall showed hyperplastic lymphoid follicles with prominent germinal centres (Figure 2). All the lymph nodes showed reactive follicular hyperplasia.

DISCUSSION

During the course of evolution of organisms, appropriate cell control mechanisms have been developed for the maintenance of proper cell numbers, differentiation, migration and adhesion. These mechanisms include signal transfer pathways, such as bone morphogenetic proteins (BMP), transforming growth factor β (TGF-β), Wnt (wingless in Drosophila), Hedge Hog and Notch. An aberration in signal pathway controlling the structure of intestinal mucosa leads to the occurrence of hamartomatous polyps which are described as "incorrectly set" proper cells.[4] The development of juvenile polyposis is associated with mutations in one of the two genes connected with TGF-β/BMP signal pathway: BMPR1A and SMAD4 with a frequency of 20% each in JPS.[5]

Hamartomatous polyposis syndromes include Peutz–Jeghers syndrome, JPS, Cowden's syndrome and Ruvalcaba–Myhre–Smith syndrome. The JPS was first described by McColl in 1964 and the minimum number of polyps required for the diagnosis varies in literature. The most widely accepted diagnostic criteria of which at least one should be present for the diagnosis of JPS are: 3-10 polyps in the colon detected on colonoscopy, juvenile polyps throughout the gastrointestinal tract or, any number of juvenile polyps with a positive family history. Solitary juvenile polyp of the rectosigmoid region occurring in 1% of children between 4 -5 years in which dysplastic changes are very rare is a separate entity from JPS. The sporadic type JPS is more common, occurring in 66% and 33% have a positive family history.[6] In a study 85% cases of JPS were diagnosed in the first and second decade of life; 98% patients had polyps in colon, 14% in stomach, 2% in duodenum and 7% in jejunum and ileum.[7] Juvenile polyps range from few millimeters to 3 centimetres in diameter, covered by columnar epithelium having mucus filled glands in an abundant lamina propria having inflammatory cells. Juvenile polyps do not have smooth muscle component which distinguishes them from Peutz–Jeghers polyps.[8]

Polyps larger than 3 cm are usually pedunculated, with mild to moderate epithelial dysplasia and have higher risk of developing adenomatous change and adenocarcinoma.[9] Patients with JPS have a higher risk of developing colorectal cancer with an incidence of 20.7% with mean age of 34 years and a cumulative cancer risk of 68% by 60 years of age.[6] The pathogenesis of carcinomatous change in hamartomatous polyps is elucidated unlike that of adenomatous polyps in which malignant transformation progresses through the adenoma-

carcinoma sequence via gatekeeper/caretaker defect. The hypothesis proposed for hamartoma-carcinoma sequence is a landscaper defect in which stromal elements alter the local environment and promote epithelial dysplasia and carcinoma.[1]

The standard methods of screening used are colonoscopy and esophagogastroduodenoscopy. The treatment depends on the clinical presentation, location and number of polyps. Surgery ranges from endoscopic removal of polyps followed by yearly surveillance to laparotomy for performing enterotomies and polypectomies. Patients with numerous colonic polyps having anaemia from excessive bleeding or failure to thrive benefit from proctocolectomy with ileorectal or ileo-anal anastomosis.[8]

Fewer patients with JPS suffer from extraintestinal congenital abnormalities like macroencephaly, hypertelorism, Meckel's diverticulum, malrotation of small bowel, mesenteric lymphangioma, undescended testes and acute porphyria.[1,3] There is a long list of anomalies such as renal agenesis, bifid uterus, atrial septal defects, arteriovenous malformations of lung, pulmonary stenosis, tetralogy of Fallot, coarctation of aorta, patent ductus arteriosus, aortic stenosis, osteoma, pectus excavatum, hereditary telangiectasia, congenital lymphedema, thyroglossal duct cyst, and amyotonia congenita.[10]

CONCLUSION

A patient with JPS, especially the sporadic type can present with problems due to extraintestinal abnormalities earlier than the symptoms of intestinal polyps. Adenomatous and dysplastic change noticed in polyps reiterates the significance of proper screening and management as there is an increased risk of malignancy.

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REFERENCES

1. Manfredi M. Hereditary Hamartomatous Polyposis Syndromes: Understanding the Disease Risks As Children Reach Adulthood. Gastroenterology & Hepatology. 2010;6:185-96.
2. Sachatello CR. Polypoid diseases of the gastrointestinal tract. J Ky Med Assoc. 1972;70:540-4.
3. Bussey HJ, Veale AM, Morson BC. Genetics of gastrointestinal polyposis. Gastroenterology. 1978;74:1325.
4. Cichy W, Klincewicz B, Plawski A. Juvenile polyposis syndrome. Arch Med Sci. 2014;10:570-7.
5. Howe JR, Roth S, Ringold JC, Summers RW, Jarvinen HJ, Sistonen P, et al. Mutations in the SMAD4/DPC4 gene in juvenile polyposis. Science. 1998;280:1086-8.
6. Jass JR, Williams CB, Bussey HJ, Morson BC. Juvenile polyposis-a precancerouscondition. Histopathology. 1988;13:619-30.
7. Desai DC, Neale KF, Talbot IC, Hodgson SV, Phillips RK. Juvenile polyposis. Br J Surg. 1995;82:14-7.
8. Calva D, Howe JR. Hamartomatous polyposis syndromes. Surg Clin NorthAm. 2008;88:779-17.
9. Subramony C, Scott-Conner CE, Skelton D, Hall TJ. Familial juvenile polyposis, study of a kindred: Evolution of polyps and relationship to gastrointestinal carcinoma. Am J Clin Pathol. 1994;102:91-7.
10. Coburn MC, Pricolo VE, DeLuca FG, Bland KI. Malignant potential in intestinal juvenile polyposis syndromes. Ann Surg Oncol. 1995;2:386-91.

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