International Journal of Science and Research (IJSR)
ISSN (Online): 2319-7064
Index Copernicus Value (2013): 6.14 | Impact Factor (2013): 4.438

Recurrent Juvenile Polyposis – A Case Report

Dr. M. Devojee[1], Dr. P. Geetha Vani[2], Dr. K. Jagan Mohan Rao[3]

[1]Associate Professor, Department of Pathology, Siddhartha Medical College, Gunadala, Vijayawada, Andhra Pradesh, PIN.520008, India

[2]Senior Resident, Department of Pathology, Siddhartha Medical College, India

[3]Consultant Gastroenterologist, Nagarjuna Hospital, Vijayawada, India

Abstract: ***Juvenile polyposis is a rare autosomal dominant condition. Hamartomatous polyps can effect entire gastrointestinal tract but usually predominate in colon and rectum. In this case report we present a case of recurrent juvenile polyposis under continuous follow up and treatment since eight years.***

Keywords: Juvenile polyposis, hamartomatous polyp, recurrent polyposis, young patient, colorectum.

1. Introduction

Juvenile polyposis is a familial cancer syndrome with autosomal dominant trait characterised by multiple juvenile polyps of gastrointestinal tract, involving predominantly the colorectum.

The first juvenile polyposis describing the histological analysis of a 30 month old child's rectal polyps was published in 1939 (1). The name juvenile polyp was given by Horrilleno et.al in 1957(2).

The presence of multiple juvenile polyps in the gastrointestinal tract was first reported in 1964 (3). Polyps are commonly known to occur in the large intestine and rectum, and may also appear in the stomach and small intestine (4)

Recent studies on juvenile polyposis syndrome have indicated a higher risk or an early malignant transformation by the age of 30 – 35yrs (5). In addition to colorectal cancer these patients carry an increased risk for the development of tumor in the stomach, duodenum, biliary tree and pancreas.

Germline mutations in SMAD4/DPC4 tumor suppressor gene account for some the cases (6). SMAD4 maps to chromosome 18q21.1, a region that is often deleted in colorectal carcinoma (7).

2. Case Presentation

The case reported here concerns a young boy 16years age, born of nonconsanguinous marriage, the eldest in the family of two children. Eight years earlier he presented with rectal bleeding accompanied by significant weight loss. Clinical examination revealed that the patient was in a poor general health and was anaemic with a Hb of 9g/dl. He underwent upper GI endoscopy and colonoscopy. Colonoscopy revealed multiple soft, mucoid and pedunculated polyps of variable size ranging from 5 to 10mm, in the rectum and distal sigmoid colon (Figure 1). Therapeutic colonoscopic polypectomy of all the polyps was done.

Grossly, the polyps were around 1cm with smooth external surface and with grey white, firm cut section(Figure2)

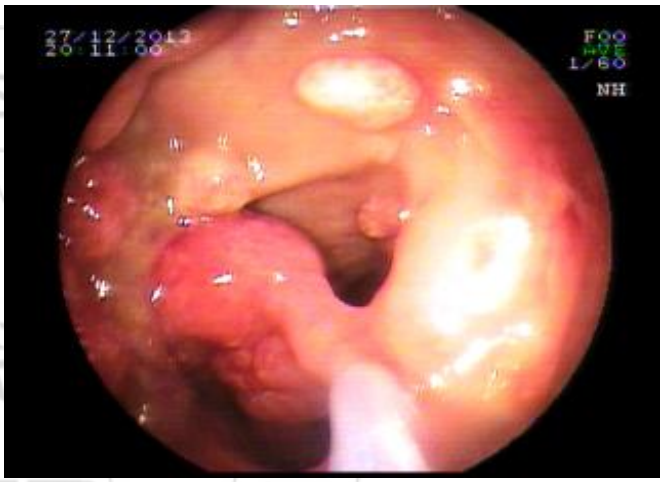

Figure 1: Colonoscopy showing multiple polyps

Figure 2 : Gross image of polyps

Histopathology revealed the presence of typical hamartomatous nonadenomatous polyps of juvenile polyposis with no areas of dysplasia(Figure 3,4).

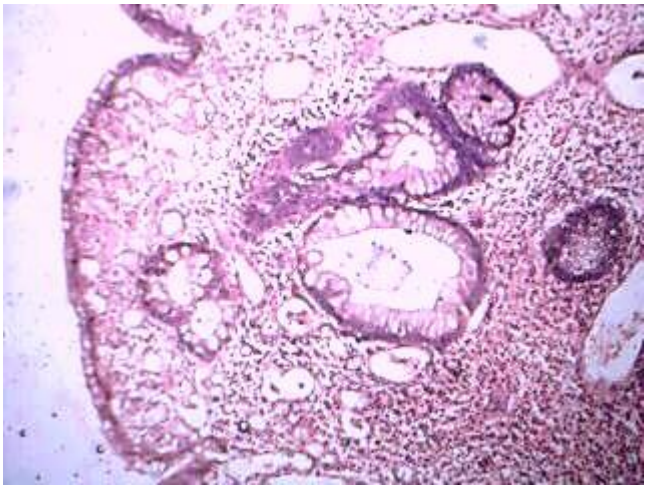

Figure 3 : Microscopy scanner view

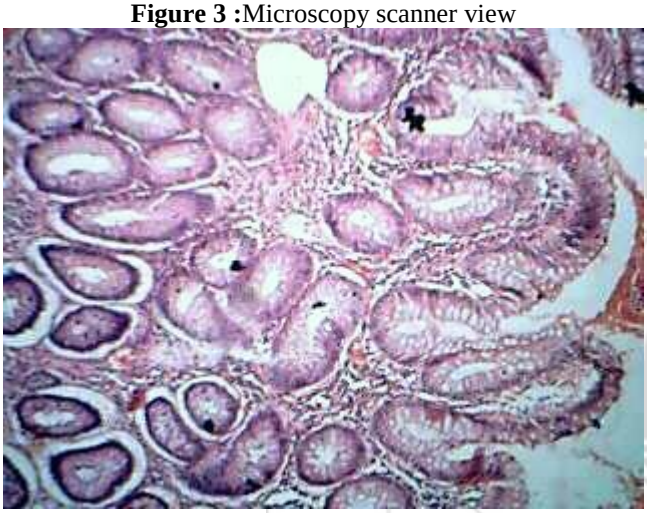

Figure 4: Microscopy low power view

There is no family history and his brother also underwent colonoscopy as per screening protocol and showed no evidence of polyps. The patient had no extra intestinal manifestations. Patient was advised follow up on a regular basis. Patient presented with recurrence in almost every follow up and underwent colonoscopic polypectomy. Upper gastrointestinal endoscopy was normal in all the follow ups. Histopathological findings remained the same, consistent with hamartomatous nature of the polyp. At present the age of the patient is 16yrs and nearly a total of 50 polyps have been excised in all follow ups.

3. Discussion

Juvenile polyposis is a rare autosomal dominant hereditary disorder characterised by multiple hamartomatous polyps most common in colon and rectum. Incidence of this syndrome is 0.6 to 1 case per 1,00,000 (8).
Following the initial report of Stamper in 1975 (9), the following diagnostic criteria are established by WHO :
1. More than 5 juvenile polyps of the colorectum.
2. Juvenile polyps throughout the gastrointestinal tract
3. Any number of juvenile polyps with a family history of juvenile polyposis.

In our case patient presented with 6 to 7 polyps in rectum and sigmoid colon. Two third of patients present within the first two decades of life with a mean age at diagnosis being 18.5 years (10). It is often established after gastrointestinal exploration for rectal bleeding, iron deficiency anaemia and chronic diarrhoea (11). Our patient was of 8 years age when he first presented with rectal bleeding.

Extra intestinal manifestations have been reported in 11 to 15% of juvenile polyposis (12) which includes congenital anomalies of heart, pulmonary arteriovenous malformations and hypertrophic osteorthopathy. Our patient presented with none.

Polyps have macroscopic and histological aspects which usually correspond to those of solitary juvenile polyp and are rounded, smooth surface with a short pedicle. Histologically, the lamina propria is abundant without smooth muscle. These polyps may be seat for focus of dysplasia. In our patient histology showed neither a contingent of adenomas or dysplasia.

For many years hamartomatous polyps were considered benign. The risk of malignancy was first described in 1966.this risk is estimated at 50% for colon and rectum , 15% for stomach,2% for pancreas and 1% for duodenum. Hence follow with colonoscopy is mandatory in all cases of juvenile polyposis. Our patient is on regular follow up for every 6 months.

4. Conclusion

Juvenile polyposis syndrome is an uncommon disease with potential recurrence and definite malignant potential with a need for prophylactic polypectomy and regular follow up of patients along with active surveillance for family members.

References

[1] Diamond M. Adenoma of the rectum in children: Report of a case in a thirty month old girl. Am J Dis Child 1939;57:360-7.
[2] Horrilleno EG, Eckert C, Ackerman LV. Polyps of the rectum and colon in children. Cancer 1957;10:1210-20.
[3] Mccoll I, Busxey HJ, Veale AM, et al. Juvenile polyposis coli. Proc R Soc Med 1964;57:896-7.
[4] Howe J. Juvenile Polyposis Syndrome. In: Scriver CR, Sly WS, Valle D, et al. eds. The Metabolic and Molecular Bases of Inherited Disease. New York: McGraw-Hill, 2001:805-818.
[5] Coburn MC, Pricolo VE, DeLuca FG, et al. Malignant potential in intestinal juvenile polyposis syndromes. Ann Surg Oncol 1995;2:386-91.
[6] 6.Houlston R, Bevan S, Williams A,Young J, Dunlop M, Rozen P, Eng C, MarkieD, Woodford RK, Rodriguez BM, Leggett B,Neale K, Phillips R, Sheridan E, Hodgson S,Iwama T, Eccles D, Bodmer W, Tomlinson I(1998). Mutations in DPC4 (SMAD4) causejuvenile polyposis syndrome, but onlyaccount for a minority of cases. Hum MolGenet 7: 1907-1912.
[7] Hahn SA, Schutte M, Hoque AT,Moskaluk CA, da-Costa LT, Rozenblum E,Weinstein CL, Fischer A, Yeo CJ, HrubanRH, Kern SE (1996). DPC4, a candidatetumor suppressor gene at human chromosome18q21.1. Science 271: 350-353.
[8] Chevrel JP, Amouroux J, Gueraud JP(1975). [3 cases of familial juvenile polyposis].Chirurgie 101: 708-721.

[9] Stemper TJ, Kent TH, Summers RW(1975). Juvenile polyposis and gastrointestinalcarcinoma. A study of a kindred.Ann Intern Med 83: 639-646.
[10] Coburn MC, Pricolo VE, DeLuca FG,Bland KI (1995). Malignant potential inintestinal juvenile polyposis syndromes.Ann Surg Oncol 2: 386-391.
[11] Brosens, L.A., van Hattem, W.A., Jansen, M., de Leng, W.W., Giardiello, F.M. and Offerhaus, G.J. (2007) Gastrointestinal polyposis syndromes. Current Molecular Medicine, 7, 29-46
[12] Hofting I, Pott G, Stolte M (1993). [Thesyndrome of juvenile polyposis]. LeberMagen Darm 23: 107-2.