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CASE REPORT

Mixed Polyposis Coli: Report of a Rare Entity with Review of Literature

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ABSTRACT

Colorectal polyps may be detected incidentally on a screening colonoscopy or when they present with symptoms like anemia or gastrointestinal bleeding. Early recognition and prompt management of polyps can cure the primary disease and prevent future risk of malignancies in the patient and provide an opportunity to screen the families in cases of inherited polyposis syndromes. We report a case of rectal bleeding due to colorectal polyps of varying histology. Histology showed hyperplastic polyp, juvenile polyps (JP) with focal dysplasia, adenomatous polyp and villous adenoma with dysplasia. He underwent total proctocolectomy with ileal pouch anal anastomosis (J-pouch) (TP-IPAA). Mixed polyposis syndrome is a rare entity. (*J Dig Endosc* 2013;4(2):39-41)

Key Words: Rectal bleeding - Hereditary mixed polyposis syndrome - Adenomatous polyps - Hyperplastic polyps - Juvenile polyps

Introduction

Colorectal polyps (sporadic or inherited) are associated with the risk of cancer either in the colon or elsewhere. The inherited polyposis syndromes are classified based on the characteristic clinical phenotype, definitive histology and the associated germline mutations of which the former two are readily available routinely in clinical practice.[1] At times, in a single patient, different polyps may show different histological subtypes or a single polyp may show multiple subtypes posing a diagnostic challenge to the clinician. This form of mixed polyposis is now increasingly being recognised as a separate entity with an entirely different phenotype and presentation.[2] We describe a patient with mixed polyposis coli who underwent TP-IPAA.

Case report

A 24 year male presented with history of bleeding per rectum for one and half years. It was small in amount, reddish in colour and mixed with stools. He was also symptomatic with generalised weakness and fatigue. There was no history of tenesmus, diarrhoea, constipation, abdominal pain or distension, vomiting or fever. There were no similar complaints or gastrointestinal malignancy in the family. Physical examination revealed anaemia with normal abdominal, per rectal and proctoscopic findings. Colonoscopy revealed multiple polyps in colon and rectum. Two large polyps in the rectum and 60 other polyps spanning the rest of the colon were removed over 4 sessions of polypectomy. The histo-pathological examination was suggestive of polyps of different varieties removed from

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colon and rectum, namely hyperplastic polyp (Figure 1), JP with focal dysplasia (Figure 2) and adenomatous polyp(Figure 3). Follow up colonoscopy revealed recurrence of polyps with an additional sessile lesion in the rectum, biopsy of which revealed villous adenoma with dysplasia(Figure 4). Barium meal follow through (BMFT) examination did not reveal any polyps in the small bowel. Contrast enhanced computerized tomography (CECT) scan of abdomen did not show any evidence of growth suggestive of colorectal malignancy, enlarged lymph node or metastasis. He underwent total proctocolectomy with ileal pouch anal anastomosis (J-pouch). Intraoperatively, there was no ascites. Small bowel, solid viscera, pelvis, peritoneum and omentum were grossly normal. The resected specimen revealed multiple pedunculated and sessile polyps throughout the colon and predominantly in rectum of various sizes(Figure 5). The patient is asymptomatic after 6 months of follow up. Family screening by flexible sigmoidoscopy did not reveal polyps in any of his first degree relatives.

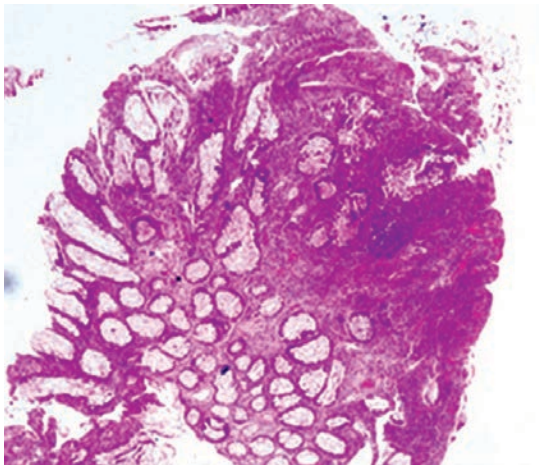

Figure 1: Hyperplastic polyp with focal ulcerations (HXE, original X10)

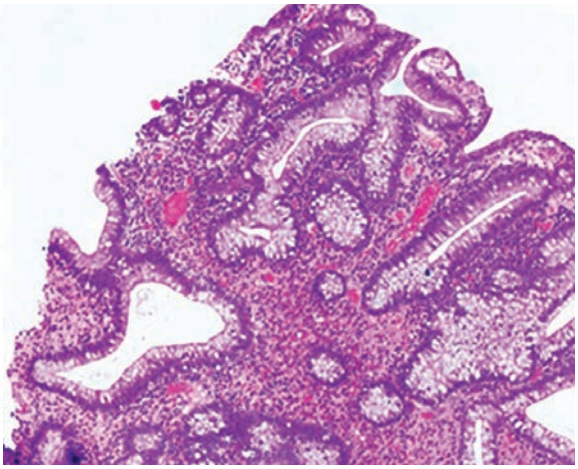

Figure 2: Juvenile polyp with smooth muscle fibres in lamina propria (HXE, original X40)

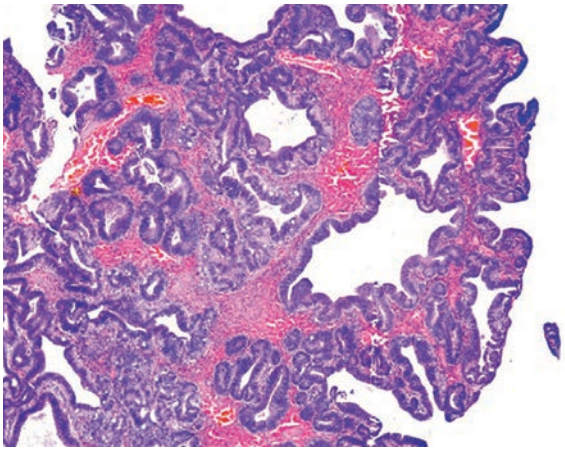

Figure 3: Adenomatous polyp with high grade dysplasia (HXE, original X10)

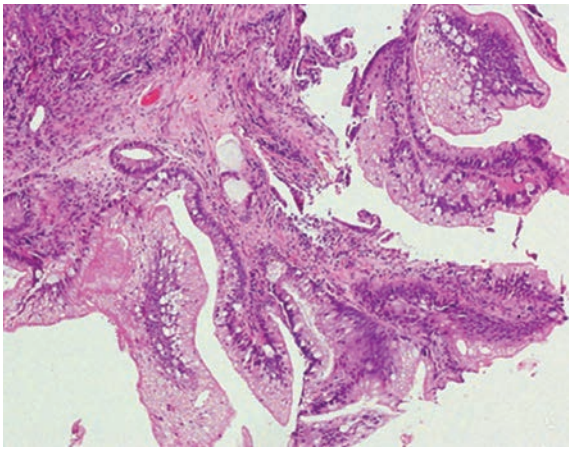

Figure 4:Villous adenoma (HXE, original X40)

Discussion

The diagnosis of polyposis coli is usually made in a symptomatic individual or during screening of an asymptomatic family member. Polyposis syndromes although rare, pose a significant clinical and genetic exercise due to the high risk of colorectal carcinoma. The commonly encountered inherited polyposis syndromes are familial adenomatous polyposis and its variants, Peutz-Jeghers syndrome and juvenile polyposis. They are easily distinguished by the characteristic phenotype and typical histology. But rarely, we see association of adenomatous polyps with JP,[3] Peutz-Jegher's polyps[4] and hyperplastic elements[5] within the same polyp or in a synchronous/metachronous lesion. The risk of colorectal neoplasia in mixed large bowel polyps was described in 1989.[6] But the hereditary nature of this mixed polyposis has been unfolded by Whitelaw *et al* in a large family (St. Mark's family 96).[7] In

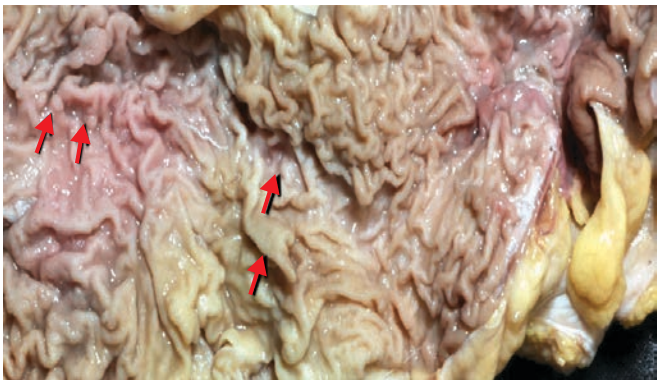

Figure 5: Resected colonic specimen showing multiple sessile polyps

hereditary mixed polyposis syndrome (HMPS) there is presence of multiple (usually <15 on initial examination) colorectal polyps of different histologies with a high likelihood of colorectal neoplasia. It apparently shows an autosomal dominant inheritance pattern and there is no risk of extracolonic malignancy unlike other inherited polyposis syndromes. The average age of presentation (half of them with rectal bleeding) is 28 years, with no evidence of gastric polyps. The average number of polyps is 1-23 which most commonly showed mixed JP and they rarely demonstrated high grade dysplasia.[2] Occurrence of mixed polyp histology in the same patient or polyp may be due to a) two or more inherited or one inherited and one acquired mutations that predispose to more than one type of polyposis, b) that all polyps may progress at different rates of through the sequence of hyperplastic→ juvenile→ Peutz-Jeghers→ adenomatous→ adenoma→ carcinoma, c) that the germline mutation may predispose to polyp formation and the histology is determined by the subsequent somatic mutations.[7] On genetic linkage analysis, the putative locus for HMPS was identified to be HMPS/CRAC1 locus on 15q13-q14 [8] thereby proving that it was not located at 6q16–61 as was previously thought.[9] Subsequently, bone morphogenesis protein receptor 1A (BMPR1A) mutations, GREM1 (bone morphogenic protein antagonist) over expression have been attributed to this disease.[10] HMPS differs from JP by the presence of lesser number of polyps (1-15), frequent adenomas and a later age of presentation. It differs from familial adenomatous polyposis coli by the presence of mixed polyposis, absence of extracolonic lesions and APC mutations. So HMPS has a distinct clinical phenotype with mixed polyp histologies as was seen in our patient. However, our case has mixed polyposis coli and we cannot label as HMPS as we couldn't perform a genetic linkage analysis and family evaluation. HMPS increases colon cancer risk, and due to low incidence of this rare disorder, screening and management guidelines are not formally made available, and standard guidelines may apply.[11] However, Whitelaw *et al* suggested a biennial colonoscopy after the age of 18 instead of a three year colonoscopic surveillance.[7]

In conclusion, mixed polyposis coli is a distinct entity of colorectal polyposis with high risk of malignancy. Identification of its existence separate from other inherited and sporadic polyposis coli is important due to the increasing recognition in the differences in the management, family screening and surveillance protocols.

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