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JUVENILE POLYPOSIS SYNDROME- AN UNUSUAL PRESENTATION

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ABSTRACT

Juvenile polyposis syndrome is characterized by multiple polyps in the colon, stomach and less commonly small bowel. Usual clinical presentation is with lower GI bleeding, mucus per rectum, abdominal pain and diarrhoea. This is a rare case of juvenile polyposis syndrome presenting with catastrophic colonic haemorrhage.

KEYWORDS: Juvenile polyposis.

CASE REPORT

A 16-year-old adolescent male presented with complaints of abdominal pain and bleeding per rectum for one month duration and aggravation of symptoms for past two days. On clinical examination, there was tachycardia with diffuse abdominal tenderness and guarding. Ultrasound abdomen showed oedematous colonic loops. A heterogeneous echotexture area with fluid-fluid level within was noted in the distal ascending colon and proximal transverse colon with no demonstrable vascularity on colour Doppler, findings indicating haematoma.

A contrast enhanced computed tomography of the abdomen along with angiogram was performed. A polypoidal submucosal lesion measuring approximately 4 x 3.5cm was noted in the proximal transverse colon with hyperdensities within. Active contrast leak was noted on arterial and venous phases. Arterial feeder to the lesion was noted from the ileocolic artery. There was an early draining vein to the superior mesenteric vein in the caecum and ascending colon. Moderate ascites was also present. Based on the computed tomography findings, a diagnosis of a submucosal polypoidal lesion with haemorrhage was made. The patient underwent explorative laparotomy and intra-operatively ascending colon, hepatic flexure and proximal transverse colon showed mucosal oedema and inflammation. Multiple clots were noted within the proximal transverse colon with dilatation of the proximal bowel loops. Pancolectomy and distal ileal resection was done. Histopathology of the resected colon showed multiple juvenile polyps, largest measuring 1 x 1 cm. Intervening mucosa was ulcerated with granulation tissue formation.

Retrospective analysis of the computed tomography images revealed multiple small polyps in the descending colon. This was a rare case of juvenile polyposis syndrome presenting with extensive bowel haemorrhage. The patient succumbed to death within a few days.

IMAGES

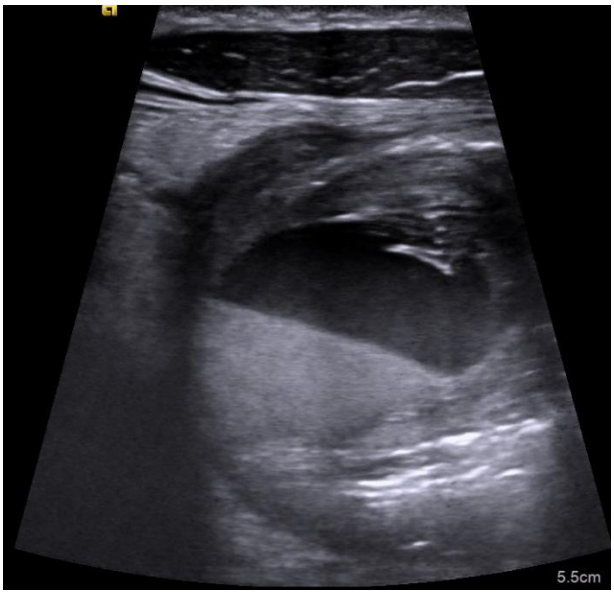


Image 1: Ultrasound image showing a heterogeneous echotexture lesion with fluid-fluid level within in the proximal transverse colon.

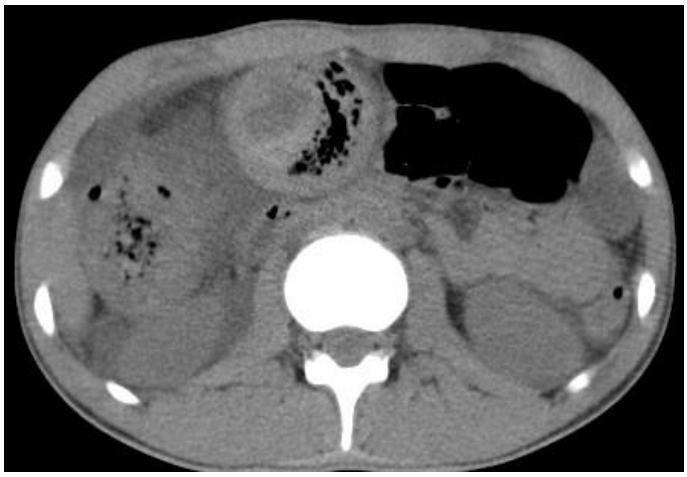
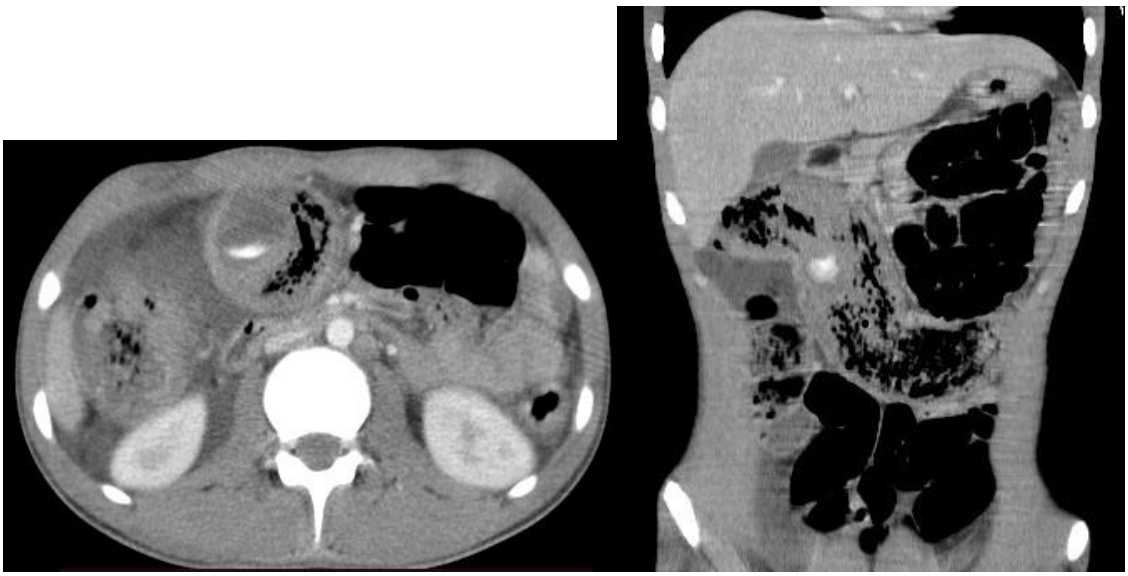
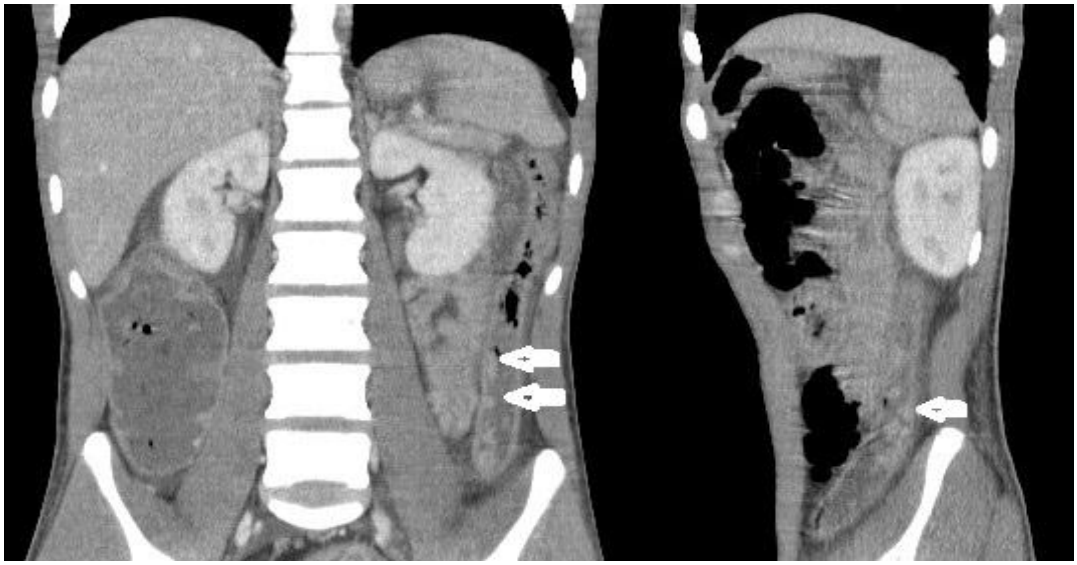


Image 2: Axial section of plain CT abdomen showing heterogeneously hyperdense lesion in the proximal transverse colon.



Images 3: a and b- Axial and coronal sections of contrast CT abdomen showing contrast leak into the lesion.



Images 4: a and b- Coronal and sagittal sections of contrast enhanced CT abdomen showing multiple small enhancing polyps in the descending colon.

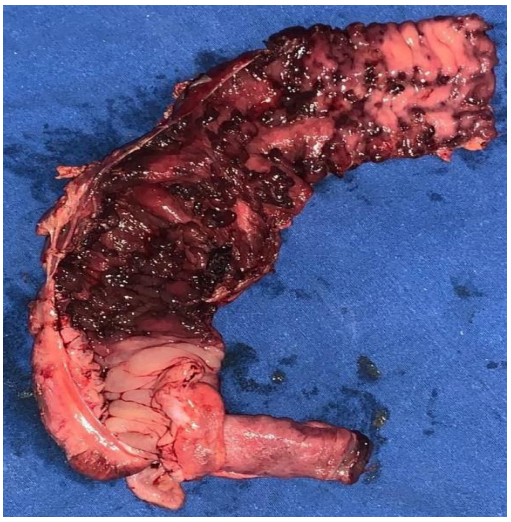


Image 5: Surgical specimen showing multiple clots within the transverse colon.

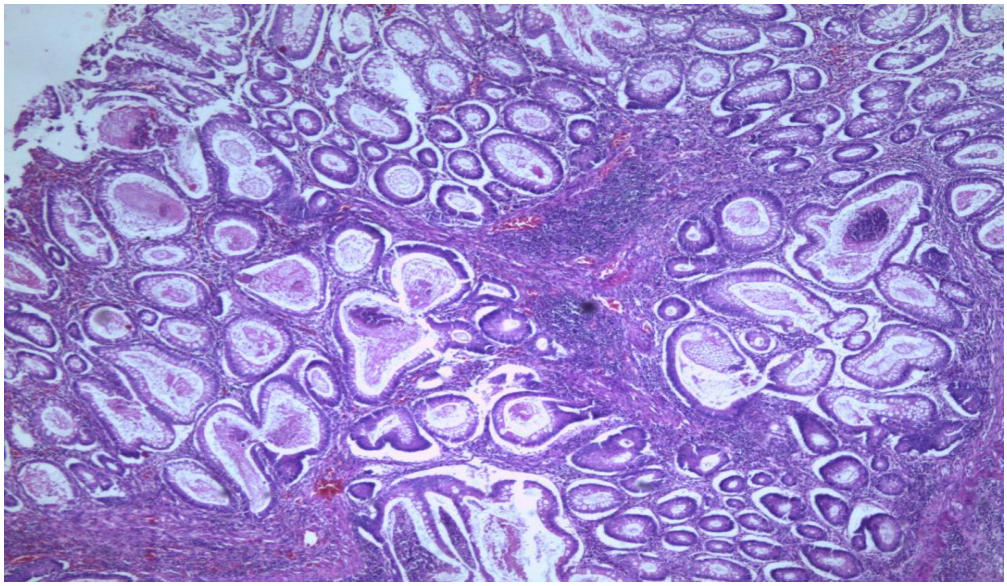


Image 6: Histopathology showing cystically dilated glands lined by columnar epithelium, containing mucin and inflammatory debris in the lumen.

DISCUSSION

Juvenile polyposis syndrome is characterized by multiple polyps in colon, stomach and less commonly small bowel. Majority of the colonic polyps are located in the proximal colon.[1]

The syndrome follows an autosomal dominant inheritance. 50 % of cases are caused by mutations in the SMAD4 gene on chromosome 10q. BMPR1A gene mutations are also reported.[2] Mutations in the DPC4 gene on chromosome 18q21.1 is also established.[6]

There are three subgroups of this syndrome based on clinical presentation: juvenile polyposis of infancy, juvenile polyposis coli and diffuse juvenile polyposis.[6]

In diffuse juvenile polyposis syndrome, the number of polyps vary from 50 to 100. These polyps are histologically composed of dilated glands filled with mucous. Inflammatory cell infiltration is often present.[3]

20% of the patients have congenital abnormalities including hydrocephalus, malrotation, cardiac lesions, mesenteric lymphangioma and Meckel's diverticulum.[2] There is increased risk of malignancy of upto 50 % with colon and stomach most commonly affected and less commonly pancreas and duodenum. The most common symptoms include lower GI bleeding, mucus per rectum, abdominal pain and diarrhoea.[4]

The methods for screening and survey of these patients include esophagogastroduodenoscopy and colonoscopy,[4] Treatment of choice is surgical colectomy with ileorectal anastomosis in symptomatic patients with endoscopic surveillance of the remaining rectum.[5]

Our case is an unusual presentation of juvenile polyps with extensive haemorrhage with multiple clots in the proximal colon resulting in mortality. The polypoidal ulceration eroding into the adjacent vessel is the possible pathogenesis for the catastrophic haemorrhage.

CONCLUSION

Juvenile polyposis syndrome could present with catastrophic colonic haemorrhage which can result in patient mortality.

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