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Case Report on Familial Adenomatous Polyposis Syndrome

Suman Choudhary[1], Dhananjay Ghodke[2]

Abstract: ***Familial adenomatous polyposis (FAP) is an inherited autosomal dominant disorder to due mutation of the tumor suppressor adenomatous polyposis coli (APC) gene located on chromosome 5q21-2. FAP is characterised by numerous intestinal adenomatous polyps, most commonly affecting large bowel. Most of the patients have family history. We reported a case of 36 years old female, who presented with acute abdominal pain, vomiting and loose stools and multiple previous hospitalisations with similar complaints. Her elder sister use to have similar complaints and was expired at young age. On evaluating the patient, she had multiple colonic polyps, ascites, dilated biliary system and pancreatitis. Her haemoglobin levels are decreasing and stool for occult is positive. Colonoscopy with biopsy is planned as largest polyp was around 2 cm so risk of malignancy is high. Total colectomy or proctocolectomy with ileoanal anastomosis is considered. More research is needed to understand the associations of FAP and their pathophysiology and guidelines for screening and treatment for this condition.***

Keywords: Familial adenomatous polyposis, FAP, APC gene mutation, colonic polyps, total colectomy

1. Case Report

A 36 years old female presented with complaints of abdominal pain, vomiting and loose stools since 2 days. Patient had history of multiple previous hospitalisation for similar complaints and was diagnosed and treated as acute pancreatitis and dilated biliary system. Her elder sister had similar complaints and died at young age. On abdominal examination she had tenderness and guarding predominantly in epigastric region with hard lump was felt in epigastric region. On lab investigations her haemogram shows decreasing haemoglobin levels and raised amylase (807 s.u.) and serum lipase (1251 U/L) and white blood cells were 14 thousand. Rest of the blood investigations were within normal limits. On ultrasound examination of abdomen showed bulky and heterogeneous echotexture of pancreas, dilated biliary system with a filing polypoidal defect and few septations within the left hepatic duct and ascites. On contrast enhanced computed tomography (CECT) of abdomen and pelvis showed innumerable enhancing polyps in ascending colon, hepatic flexure, transverse colon, descending colon and proximal rectum with bulky nodular pancreas with loss of its feathery appearance. And biliary system (Including common bile duct, common hepatic duct, right & left hepatic ducts and intrahepatic biliary radicles) were dilated with a polypoidal lesion in second part of duodenum. Additionally, there was picture of CT hypoperfusion syndrome including collapsed inferior vena cava, hyper-enhancing bilateral kidneys and enhancing thickening of small bowel. There was a non-enhancing soft tissue density nodule was noted in subcutaneous plane of left gluteal region just adjacent to natal cleft and was retrospectively confirmed as epidermoid cyst on ultrasound and histopathological reports. Histopathological reports (HPRs) of biopsies from colon revealed adenomas with malignant transformation. Total proctocolectomy with ileoanal anastomosis was planned and consent for same was obtained. The children have been advised regular follow-up with annual sigmoidoscopy.

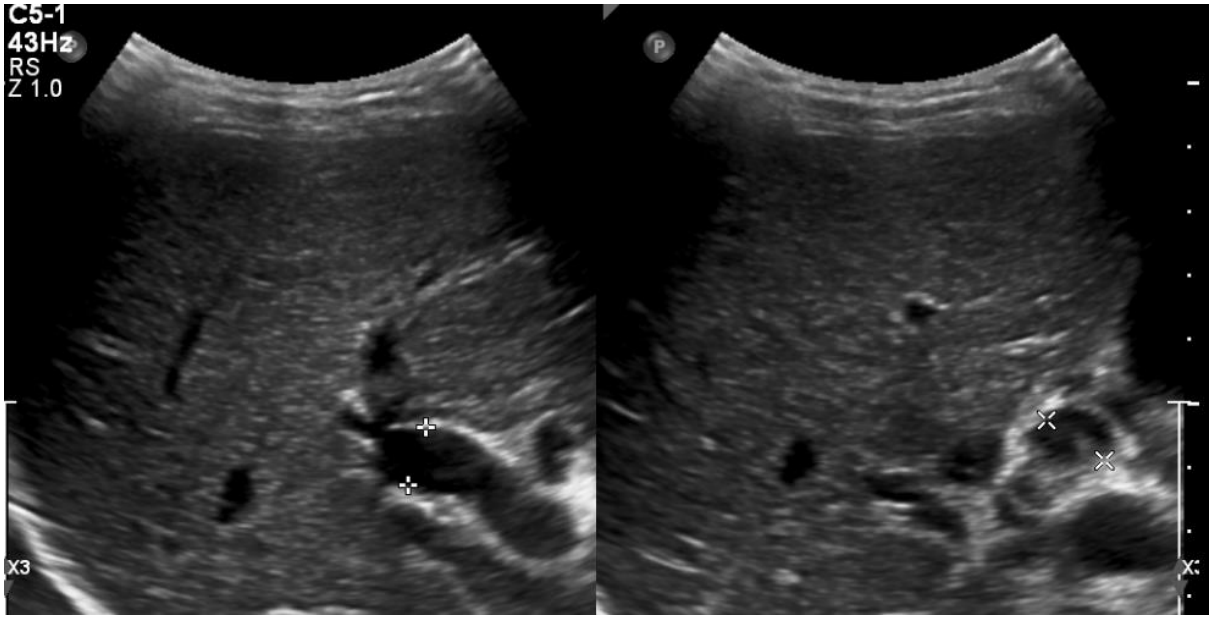

Figure 1: Ultrasound abdomen shows common dilated hepatic duct and left hepatic duct with filling defect and septations within the left hepatic duct.

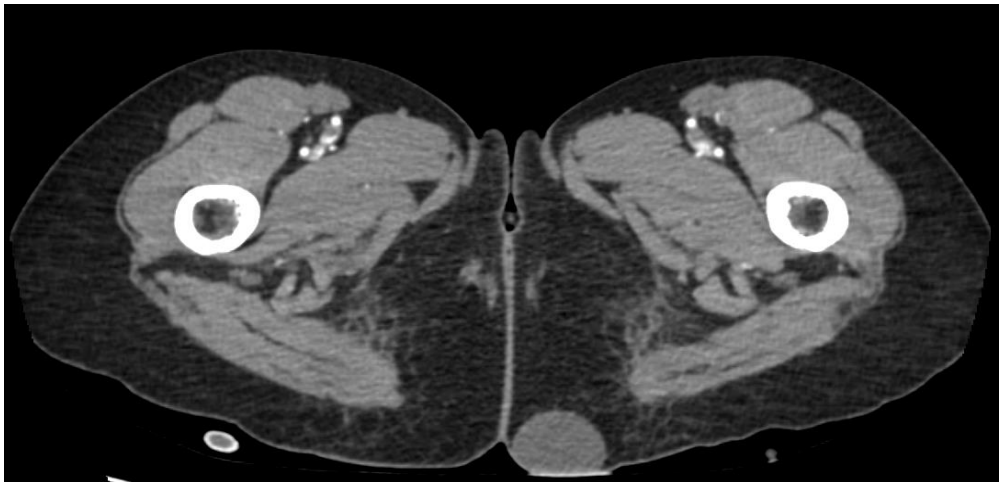

Figure 2: Contrast enhanced CT, axial view shows non-enhancing soft tissue density nodule seen on left of natal cleft.

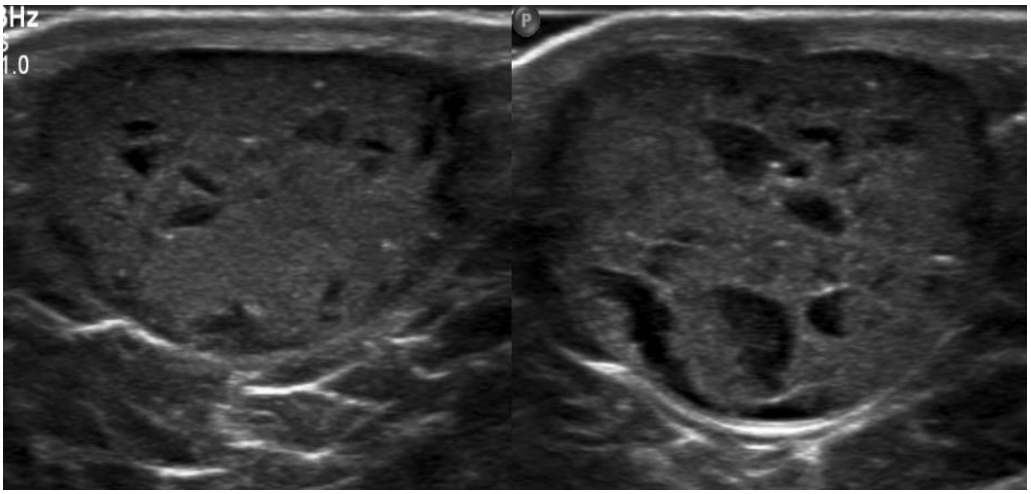

Figure 3: Local ultrasound of lesion mentioned in figure shows well defined ovoid lesion with filiform anechoic areas and linear hyperreflective disc like areas representing keratin, suggestive of epidermoid cyst.

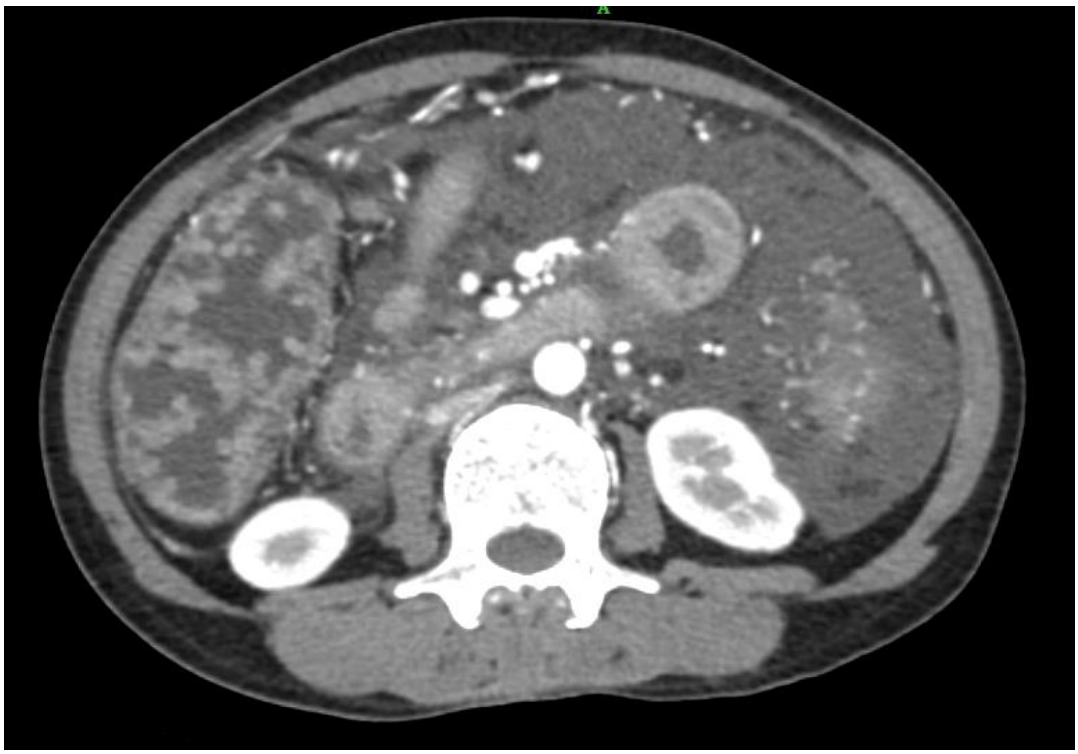

Figure 4: Contrast enhanced CT axial view showing multiple enhancing polypoidal lesions in hepatic flexure of colon and free fluid in abdomen.

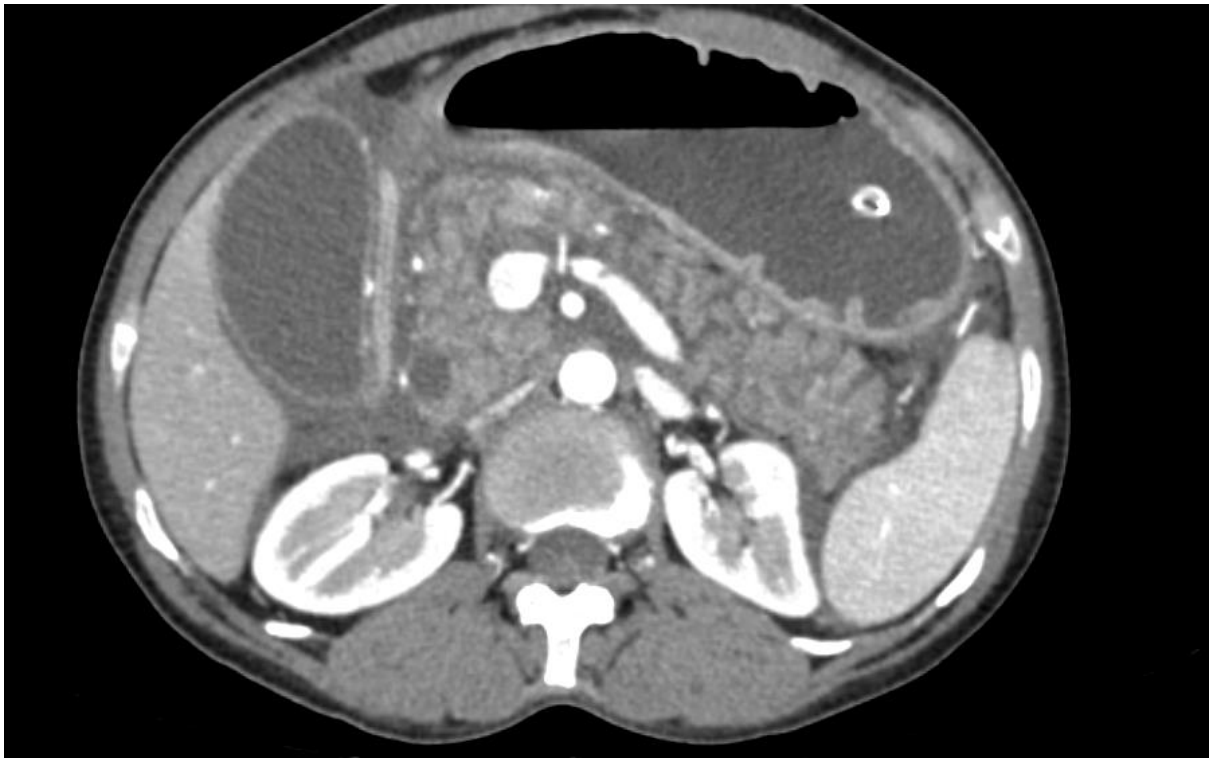

Figure 5: Contrast enhanced CT axial view showing nodular & bulky pancreas and dilated distal CBD.

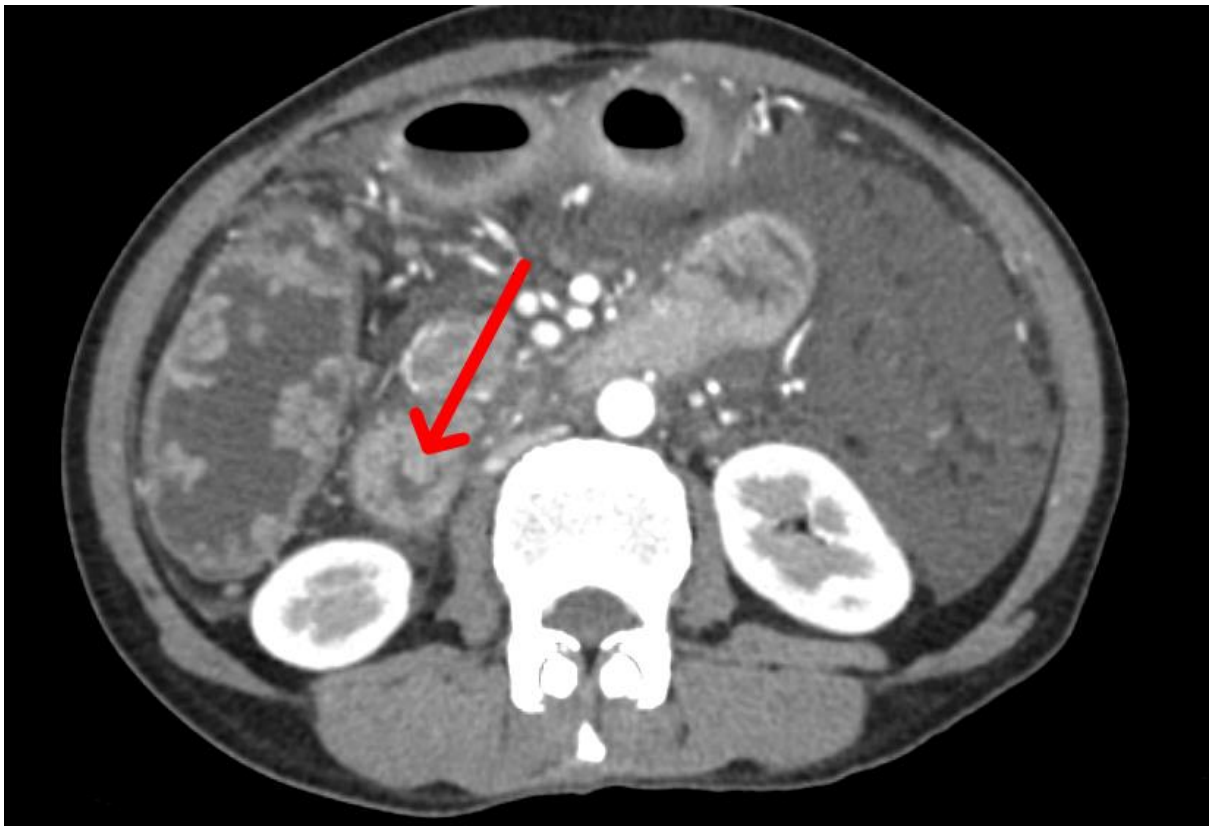

Figure 5: Contrast enhanced CT axial view shows polypoidal lesion in second part of duodenum (Red arrow)

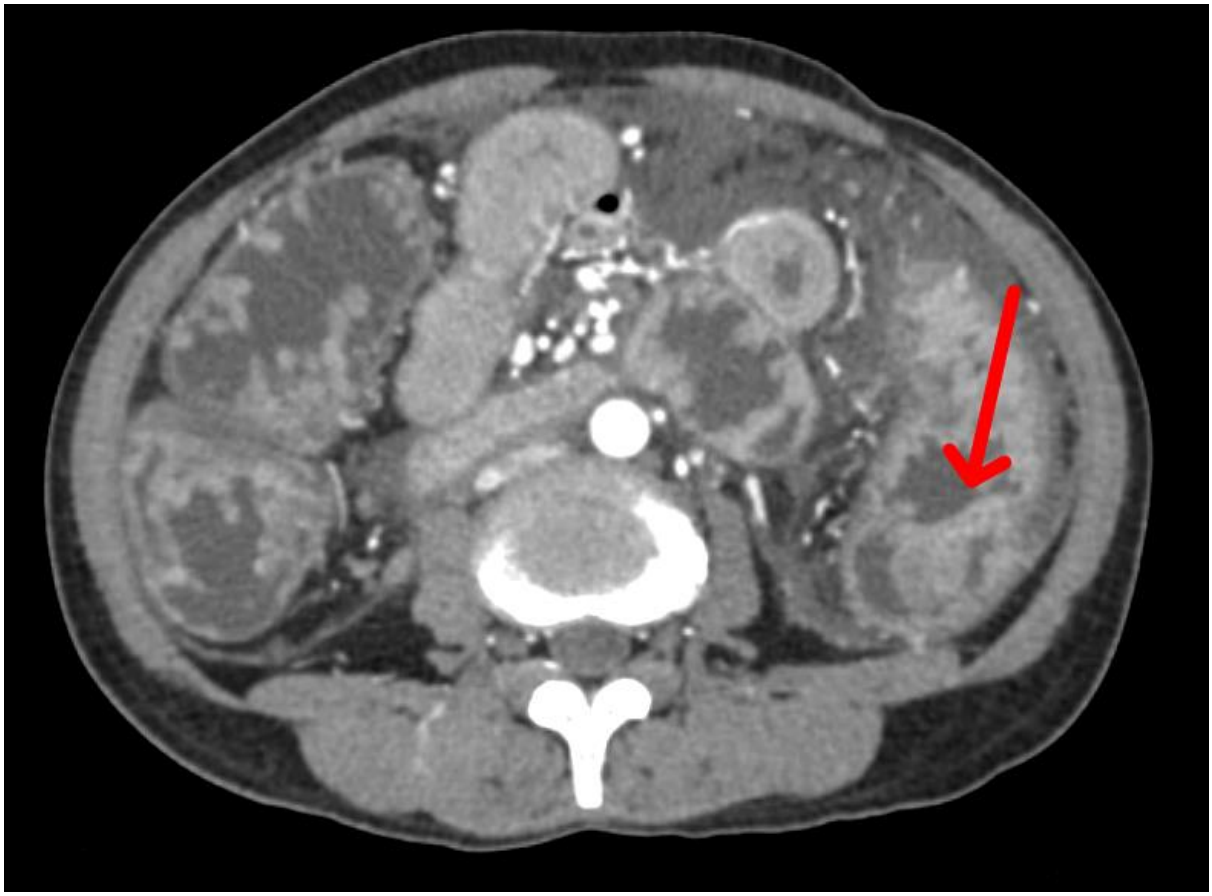

Figure 6: Contrast enhanced CT axial view showing large polypoidal lesion in proximal descending colon (Red arrow)

2. Discussion

Familial adenomatous polyposis syndrome having in numerous polyps predominantly affecting large bowel it is an autosomal dominant inheritance caused by APC gene mutations located on chromosome 5q21 [1]. Both genders are equally affected and most of the patient have family history of colorectal polyps but "de novo", without any clinical or familial history of FAP [3]. Patient's family history was significant. Other common clinical features in patients with FAP include multiple gastric polyps, duodenal & periampullar adenomas, while extra intestinal features are desmoid tumours, congenital hypertrophy of the retinal pigment epithelium (CHRPE), epidermoid cysts, osteomas and thyroid cancer [4]. This patient had epidermoid cyst. Cause for dilated biliary system is most likely due to duodenal adenomas and biliary polyps which are common association in cases of FAP. Pancreatitis and pancreatic carcinoma is also associated with it, pancreatitis may be due to duodenal adenoma, idiopathic recurrent pancreatitis [6], autoimmune or may be associated SPINK1 gene mutation. Diagnostic criteria include hundred or more colorectal adenomatous polyps or Germline mutation in APC or Family history of FAP with colorectal adenomas (age < 30) or Family history of FAP and presence of at least one epidermoid cyst, osteoma or desmoid tumor. This diagnostic criterion was fulfilled by the patient. Prophylactic surgery is recommended before the age of 25years, options are total proctocolectomy with ileostomy; (2) subtotal colectomy with ileorectal anastomosis, and (3) restorative proctocolectomy.

3. Conclusion

Patients with FAP may present with vague abdominal complaints and without any family history, hence need to be carefully evaluated. Good patient compliance is of prime importance in deciding the treatment and surveillance modality subsequently determining the prognosis of patients with FAP.

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