Incidentally detected adrenocortical carcinoma in familial adenomatous polyposis: an unusual presentation of a hereditary cancer syndrome

Samarth Agarwal, Ashish Sharma, Deepanshu Sharma, Satyanarayan Sankhwar

Department of Urology, King George's Medical University, Lucknow, Uttar Pradesh, India

Correspondence to
Dr Ashish Sharma,
ashishuc343@gmail.com

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DESCRIPTION

A 30-year-old woman presented to us with complaints of intermittent bleeding per rectum, vague abdominal pain and altered bowel habits for last 3 months. There was no history of headache, palpitations, diarrhoea, weight loss, skin lesions, haematuria and hypertension. Her grandmother had history of colostomy for acute intestinal obstruction and died of unexplained cause at age of 55 years. Her laboratory investigations were normal. On digital rectal examination, multiple polypoidal firm masses were felt approximately 1–5 cm from anal verge. On colonoscopy, number of polyps were seen in the colon around 40–50 in number, ranging from few millimetres to largest measuring around 3 cm. These polyps spread over ascending to sigmoid colon and rectum in increasing number. The biopsy from these polyps was suggestive of tubular adenomas. A definitive diagnosis of familial adenomatous polyposis (FAP) was made. FAP is associated with a myriad of presentations, and hence, a thorough work up was done to elicit associated lesions. An upper gastrointestinal endoscopy was performed, which was unremarkable. On slit-lamp examination, discrete flat multiple pigmented lesions were present in retina suggestive of congenital hypertrophy of retinal pigment epithelium (CHRPE). However, the patient did not complain of any eye symptoms.

Further evaluation with contrast-enhanced CT scan (CECT) of abdomen showed multiple enhancing polypoidal mass lesions (largest of size 3×2 cm) throughout the colon and rectum with no evidence of any regional or distant metastasis. To our surprise, CECT also revealed a heterogeneously enhancing, well-defined adrenal mass measuring 8.5×8.5×6.5 cm with areas of necrosis suggestive of localised adrenocortical carcinoma (ACC) (figure 1). Functional adrenal work-up (serum aldosterone, serum cortisol amd 24 hours urinary metanephrine and normetanephrine) results were within normal range.

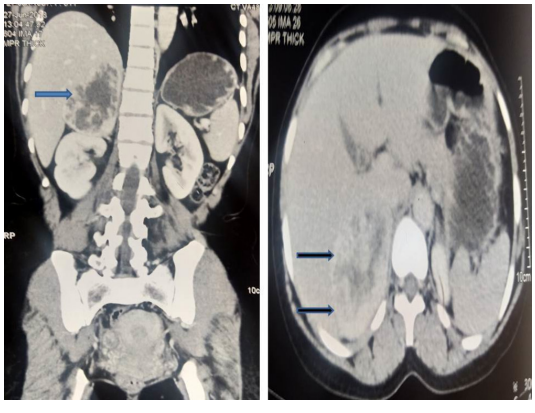

Figure 1 Contrast-enhanced CT scan abdomen (coronal and transverse sections) showing heterogeneously enhancing, well-defined adrenal mass measuring 8.5×8.5×6.5 cm with areas of necrosis suggestive of localised adrenocortical carcinoma.

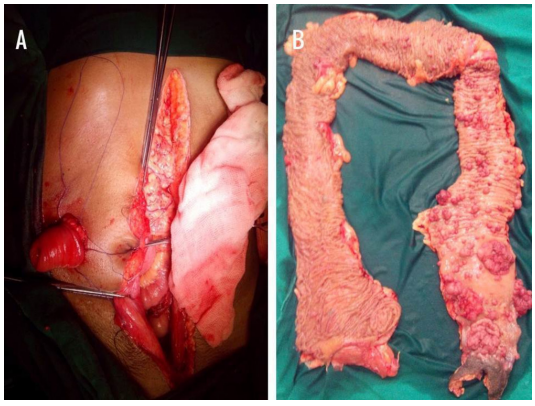

Figure 2 (A) Intraoperative picture showing total proctocoelectomy with permanent end ileostomy. (B) The total proctocoelectomy specimen showing multiple poylpoidal lesions throughout the rectum and sigmoid.

After proper counselling, she underwent radical adrenalectomy with total proctocoelectomy with permanent end ileostomy as shown in figure 2A. The total proctocoelectomy specimen was examined and revealed multiple poylpoidal lesions as shown in figure 2B. Blood sample of patient was analysed for truncated APC gene product, and in vitro protein synthesis assay was positive. Histopathology and immunohistochemistry (synaptophysin and inhibin) examination revealed high-grade ACC with negative surgical margin with pT3N0M0 staging. The postoperative period was uneventful. She was discharged on postoperative day 8. She was doing well at 4 weeks follow-up with no significant complains.

The FAP is an autosomal-dominant colorectal cancer syndrome, which is caused by mutation of APC gene on chromosome 5q21. It is characterised by occurrence of hundreds of adenomatous colorectal polyps and almost inevitable progression to colorectal cancer by the fourth decade of life. Other associated features of FAP include

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upper gastrointestinal polyps, desmoid tumours, CHRPE and other extraintestinal malignancies. Uncommonly, endocrine tumours like parathyroid, pituitary, pancreatic and adrenal neoplasms have been described in these patients.[1]

Due to widespread utilisation of modern imaging modalities like CT and MRI, the detection rates of adrenal incidentalomas has increased during the last decades. About 7% of patients with FAP or its variants have adrenal masses, as compared with 3% in general populations.[2] The biological behaviours of adrenal neoplasm in patients with FAP are comparable with sporadic variety but its required longer follow-up.[3] Mostly adrenal lesions are non-functional and benign, while functioning adrenal tumour mostly secrets cortisol. These non-functional adrenal tumours are asymptomatic and are usually detected incidentally or at autopsy. ACC is a rare neoplasm of adrenal gland with poor prognosis with incidence of one to two per million populations. The ACC may be associated with Li-Fraumeni, Lynch syndrome, Beckwith-Wideman, MEN type 1 and FAP. There are only few reported cases showing association between FAP and ACC.

Learning points

- Familial adenomatous polyposis (FAP) is an autosomal-dominant colorectal cancer syndrome, which is characterised by occurrence of hundreds of adenomatous colorectal polyps and almost inevitable progression to colorectal cancer by the fourth decade of life.
- The extraintestinal manifestation includes desmoid tumour, retinal lesions and endocrine tumours like parathyroid, pituitary, pancreatic and adrenal neoplasms.
- The clinical presentation and biological behaviours of adrenal neoplasm in FAP is comparable with sporadic cases, and same protocols are followed in these cases.
- Adrenocortical carcinoma is a rare, mostly non-functional neoplasm of adrenal gland with poor prognosis.

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