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Adrenocortical Carcinoma as an Extracolonic Manifestation of Familial Adenomatous Polyposis: A Case Report

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Abstract

Familial Adenomatous Polyposis (FAP) is an autosomal dominant inherited disease characterized by multiple colorectal polyps with a high potential to develop colorectal cancer. FAP has diverse extracolonic manifestations including adrenal masses. The prevalence of Adrenal Incidentalomas (AI) in FAP patients is 13%, 97% of which are benign. Adrenocortical carcinoma as an AI in FAP patients is very rare and limited to few case reports in the literature. We are reporting a rare case of a young male who was referred to urology for a large adrenal mass, and underwent adrenalectomy and the reported pathology was adrenocortical carcinoma.

Keywords: Familial adenomatous polyposis • Adrenocortical carcinoma • Adrenal incidentaloma • FAP • ACC

Introduction

Familial Adenomatous Polyposis (FAP) is an autosomal dominant inherited disease that is characterized by multiple colorectal polyps with a high potential to develop colorectal cancer [1]. The disease is caused by a germline mutation in the tumor suppressor gene called Adenomatous Polyposis Coli (APC) [1]. FAP has diverse extracolonic manifestations including adrenal masses. The prevalence of Adrenal Incidentalomas (AI) in FAP patients is 16% [2]. A retrospective study that included 311 patients with FAP and AI showed that 97% of the adrenal incidentalomas were benign and 80% of which were adenomas. The current literature regarding the associated adrenal masses in patients diagnosed with FAP, specifically Adrenocortical Carcinoma (ACC) is largely confined to limited case reports. Herein, we're reporting a rare case of a 16 years old male patient diagnosed with FAP and found to have a large left nonfunctioning adrenocortical carcinoma.

Case Presentation

This is a 16 years old male, medically free, he was investigated for FAP as his mother has the disease. The sigmoidoscopy showed <20 polyps, also the genetic testing was positive for APC pathogenic mutation. Initial imaging (CT scan) showed a large left adrenal mass measuring 8.4 × 8.2 × 10.6 cm which is suspicious of malignancy (Figure 1). Physical examination revealed normal vital signs, no palpable abdominal masses and the laboratory workup including endocrinologic testing were all unremarkable. The patient underwent left open adrenalectomy, total proctocolectomy, ileal pouch-anal anastomosis, and ileostomy creation. The pathology of the adrenal mass showed adrenocortical carcinoma. The pathology of the colon was negative for malignancy.

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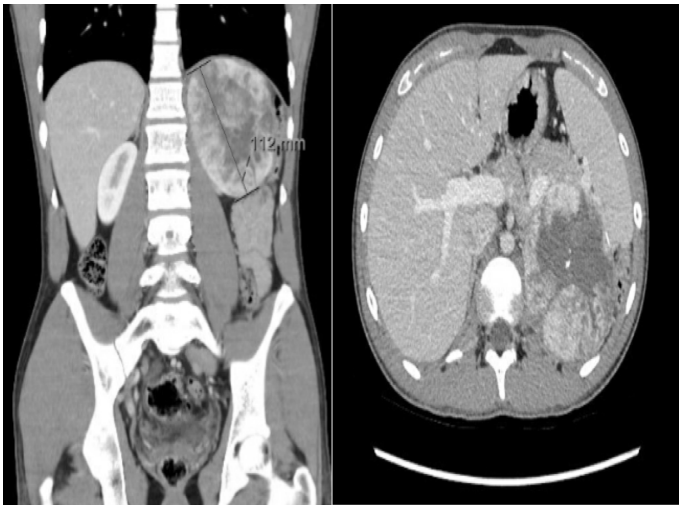

Figure 1. CT scan abdomen and pelvis with IV contrast: A large left adrenal mass measuring 8.4 × 8.2 × 10.6 cm shows a heterogeneous avid enhancement with central necrosis and few calcific foci suspicious for adrenocortical carcinoma/ pheochromocytoma.

Genetic analysis

A pathogenic variant (c.1744-2A>G) causes an A>G nucleotide substitution at the -2 position of intron 14 of the APC gene. Functional RNA studies have shown that this variant causes skipping of exon 15 and this mutant transcript is expected to create a frameshift and a premature translation stop signal and be expressed as a truncated protein.

Gross description

The specimen consists of an oval-shaped firm mass weighing 333 grams and measuring (11 × 8 × 4.5 cm). The outer surface is smooth and glistening. The opening revealed an encapsulated well-circumscribed fleshy, tan-to-brown mass measuring 11 × 8 × 4.5 cm with a heterogeneous cut surface and extensive areas of necrosis measuring (10 × 5.5 × 3 cm). The mass is entirely replacing the adrenal gland. No remnant of the normal gland.

Histopathologic findings (Figure 2)

Adrenal cortical carcinoma, oncocytic type.

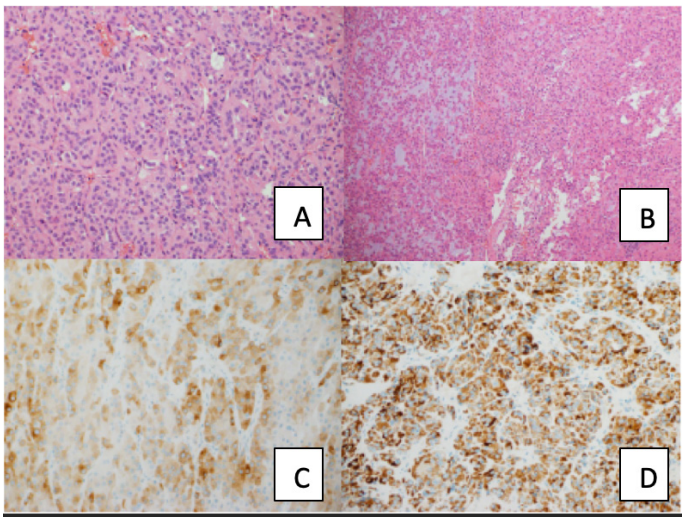

Figure 2. Pathology: **A)** H&E High power field; **B)** H&E low power field; **C)** Inhibin and **D)** Melanin A.

Mitotic count: Eleven mitosis/50 HPF, Ki67 (~50).

Extensive tumor necrosis was noted (~50%).

Positive: Melanin A, Inhibin.

Negative: Chromogranin.

Discussion

Adrenal Incidentaloma (AI) is defined as an adrenal mass that is found incidentally with a size of more than 1cm without any clinical suggestion of functionality (Hormonal production) [3]. FAP patients frequently undergo cross-sectional imaging to investigate desmoid tumors and other extracolonic manifestations. AI is common in patients with FAP with an incidence rate above 10%. The literature showed that the majority of these masses are benign in nature [2]. ACC in FAP patients is very rare and confined to very limited case reports. ACC in general is a very rare disease with an estimated prevalence of 0.72 cases per 1 million population and accounting for 0.2% of all cancer deaths in the United States [4].

AI that is found in FAP patients seems likely to mirror sporadically found adrenal lesions [3]. A prospective cohort study was conducted aiming to explore the natural history of AI in patients with FAP and the suggested management model [5]. The study included 30 patients and they've been followed with a mean period of 112 months. It showed that the majority of the AI are benign and clinically silent lesions, and those who had surgical excision were based on the initial scan features. Moreover, none of the benign adrenal lesions at the diagnosis showed aggressive growth over the follow-up period [5].

The study suggested that these patients should be managed according to initial imaging, if the size of the mass is above 3.5 cm or it has suspicious imaging characteristics (Calcification, Hounsfield >10), refer the patient to an endocrinologic/surgical clinic for further evaluation. Also, the history and the vital signs must be obtained to exclude subclinical hyper functioning mass. Blood and urine workup is also recommended to be done (Electrolyte, 24-hour acidified urine collection for free catecholamines, ACTH, U/E). If the workup is negative, it is recommended to follow the patient with imaging one year apart from the initial study to exclude rapidly growing mass. If the repeated scan showed no significant change in size or the radiological features of the mass proceed with two more MRI/CT scans, at approximately three-year intervals. As these patients frequently need scans for monitoring of desmoid disease and other FAP manifestations, the scan interval and modality can be flexible to coincide with other imaging.

Conclusion

For the patient in our case report, the initial imaging showed a large mass that was above 3.5 cm which has suspicious imaging characteristics (Central necrosis and peripheral calcifications). The patient was referred to endocrinology for further testing to rule out subclinical hyperfunctioning mass/pheochromocytoma. The workup came back negative for which the patient was planned for surgical excision. Despite the low incidence of such disease (ACC) in the FAP population, careful follow-up is recommended to avoid dramatic consequences and to plan intervention promptly.

Ethical Statement

Informed consent was obtained from the patient for writing and publishing this case report and any accompanying images.

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Declaration of Interest

The authors have no conflicts of interest.

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