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Case Report

An uncommon intersection: Familial adenomatous polyposis and solid pseudopapillary neoplasm of the pancreas: A case report and review of the literature ☆,☆☆

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ABSTRACT

The co-occurrence of both Familial Adenomatous Polyposis (FAP) and Solid Pseudopapillary Neoplasms (SPN) of the pancreas is extremely uncommon, with limited reports published in the literature. FAP is a rare inherited disorder caused by a mutation in the adenomatous polyposis coli (APC) gene, while SPN is generally a low-grade malignant pancreatic lesion. We present the case of a 33-year-old female with a familial history of FAP, who initially presented with breast fibromatoses and was subsequently found to have colonic polyps, consistent with FAP, along with rare events like pancreatic SPNs in the head and tail of the pancreas and large desmoid tumors. It is a unique case that has never been reported in the literature and we provide findings of computed tomography (CT) and volume rendering to correlate the radiological features with pathology for an optimized diagnosis.

Introduction

Familial adenomatous polyposis (FAP) is an autosomal dominant syndrome, with both intra-colonic and extracolonic manifestations [1]. Solid Pseudopapillary epithelial neoplasms (SPN) are low-grade malignant neoplasms, classically occurring in young females, constituting, 0.17%-2.7% of pancreatic tumors [2]. FAP usually has a mutation in the adenomatous polyposis coli (APC) gene, which activates the downstream Wnt/ β-catenin signaling pathway, which is thought to be the reason for SPNs in such patients. FAP and SPN occurring together is a rare combination, and only 7 cases have been reported in the literature, however, none of them had multiple SPNs. We report a case of a young female with multiple extracolonic manifestations of FAP including desmoid tumors, and multifocal SPNs.

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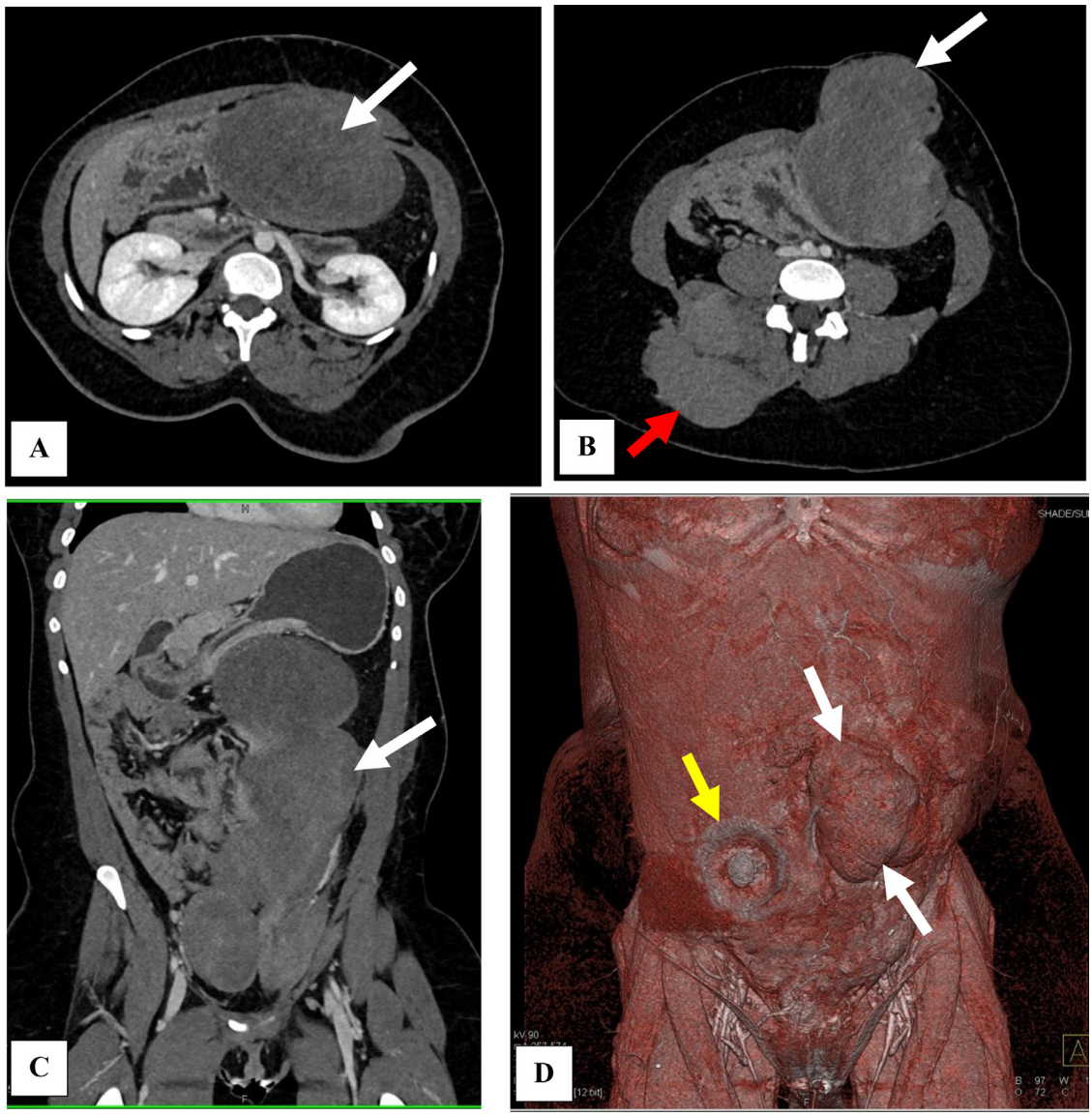

Fig. 1 – A 33-year-old female with a history of FAP and multiple desmoid tumors. (A-C) A large subcutaneous and intramuscular lesion within the left abdominal wall, extending vertically down the left abdomen seen in both axial and coronal CT scans, measuring approximately 12.5 cm x 11.1 cm in the largest diameter, exerting mass effect on nearby structures (white arrow). Another large lesion in the paraspinal region measuring 7.7 x 4.3 cm (red arrow). They are likely to be desmoid tumors, consistent with the patient's history. D: 3-dimensional volume rendering showing in detail the growth and extent of a large desmoid tumor in the left abdominal wall (white arrow). An ileostomy stoma can be seen on the right side (yellow arrow) as the patient underwent proctocolectomy and diverting ileostomy for FAP.

Case presentation

A 33-year-old female, with a history of Familial Adenomatous Polyposis, and desmoid tumors, was noted to have masses in the pancreatic head and tail.

At initial presentation, she was noted to have a large non-tender mass in the right breast. She had a past medical history of asthma, postpartum depression and a heart murmur and a past surgical history of appendectomy. She also reported a strong oncologic family history of colon, liver, ovarian and gastric cancers. She underwent resection of the breast mass, which was diagnosed as fibromatoses by pathology. Later, she developed multiple masses in her left anterior abdominal wall, left chest wall, and right paraspinal soft tissues. She underwent multiple resections of these masses which were consistent with desmoid tumors. However, they regrew and continued to increase in size, exerting an extrinsic mass effect on nearby structures, very well visualized on CT 3-dimensional volume rendering (Fig. 1). She was treated with a regimen of Sorafenib and Doxil.

She had intermittent GI symptoms including blood in stool, constipation, abdominal pain, and bloating, and was scheduled for colonoscopy. Due to a strong maternal family history

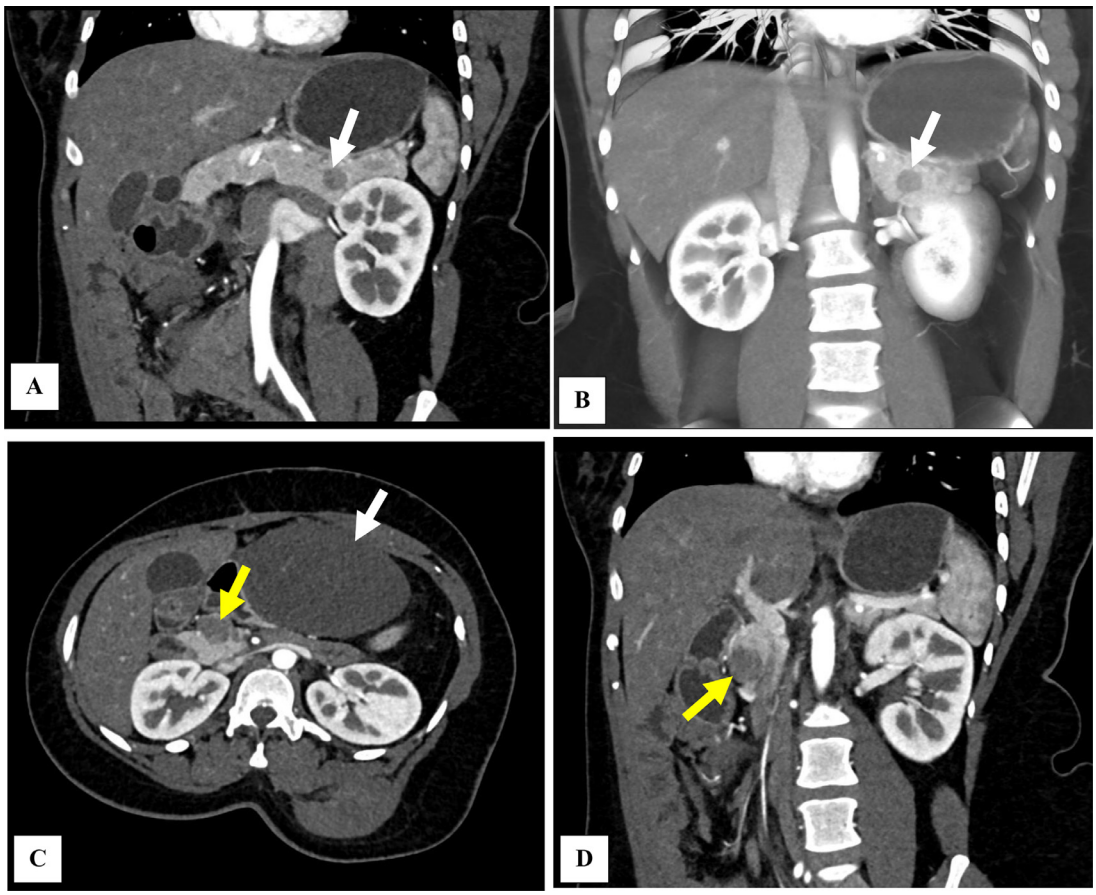

Fig. 2 – A 33-year-old female with a history of FAP and multiple desmoid tumors. (A and B) Contrast-enhanced CT scans with coronal (A) and volume rendering (B) images showing a mass in the pancreatic tail, approximately 1.4 cm (white arrow). (C) Contrast-enhanced axial CT scan demonstrating a large left abdominal wall mass, likely a desmoid tumor, compressing the bowel loops (white arrow). A lesion is also seen in the pancreatic head (yellow arrow). (D) Coronal CT scan again demonstrating the pancreatic head lesion, approximately 2.3 cm in diameter (yellow arrow).

of polyposis and colon cancer, she underwent genetic testing. Analysis of the APC gene revealed that she has a deletion of exons 6-16, confirming the diagnosis of familial adenomatous polyposis (FAP). Multiple sessile and pedunculated polyps were identified on colonoscopy, extending down to the dentate line, histologically proven to be tubular adenomas. She underwent open restorative proctocolectomy with mucosectomy, ileal pouch-anal anastomosis, and diverting loop ileostomy.

Later, she presented with abdominal distension, pain, and nausea. Upon examination of the surgical stoma, it appeared edematous with decreased output. Although she was managed symptomatically, the abdominal pain continued. A CT scan was conducted to further evaluate her symptoms. The scan revealed a mass in the head of the pancreas, which was also noted in scans taken a year prior. Additionally, a second mass was identified in the tail of the pancreas (Fig. 2). An endoscopic ultrasound with fine needle aspiration (EUS/FNA) was performed on both masses. Histological analysis confirmed that they were multifocal solid pseudopapillary neoplasms, which tested positive for nuclear beta-catenin, progesterone receptor (PR), and CD10, and exhibited a low Ki67 index. The patient was experiencing uncontrolled abdominal pain due to multiple desmoid tumors and was followed by a pain specialist for palliative care. Due to multiple comorbidities and risk of further deterioration after surgery, the patient was kept on active surveillance for SPN.

Discussion

Familial adenomatous polyposis (FAP) is an autosomal dominant syndrome with a germline mutation in the adenomatous polyposis coli (APC) tumor suppressor gene [3]. It causes abundant adenomatous polyps and inevitably develops into colorectal cancer if not removed [4]. It has several extraintestinal manifestations including desmoid tumors, fibromas, multiple osteomas, dental abnormalities, brain tumors, and other lesions throughout the body [1].

A common benign extracolonic manifestation of FAP is desmoid tumors. These are slow-growing fibroblastic tumors that display aggressive local behavior, often infiltrating surrounding structures and resulting in a high rate of local re-

Table 1 – Review of reported cases in the literature of SPN and FAP occurring together.

Author	Age	Gender	Location of SPN	Size of SPN	Genetic mutation in SPN	APC Mutation	Management	Survival	F/U time
Ruo et al. [13]	43	Female	ND	ND	ND	ND	PD	Death due to colorectal cancer metastasis	71 months
Farahmand et al. [14]	19	Female	Head	ND	CD10, synaptophysin, chromogranin A, vimentin, neuron-specific enolase (NSE)	+	Resected	Alive – no recurrence	18 months
Inoue et al. [15]	30s	Male	Head	12 × 10 mm	CD10, vimentin and B-catenin	ND	Resected	ND	ND
Naoi et al. [16]	20	Female	Tail	10 cm	B- catenin	+	Resected	Alive – no recurrence	5 years
El Halabi et al. [17]	25	Female	Head	7 × 6.3 × 7 cm	B-catenin, CD-10	+	PD	Alive – no recurrence	2 years
Meira-Junior et al. [18]	54	Male	Tail	1.2 cm	cytokeratin, vimentin, progesterone receptor, cyclin D1, CD99, beta-catenin	ND	Distal pancreatectomy with splenic preservation	Alive – no recurrence	ND
	34	Female	Neck	2.4 cm	beta-catenin	ND	Central pancreatectomy with Roux-en-Y pancreatojejunostomy	Alive – no recurrence	ND
Arshad et al. 2024 (this study)	33	Female	Head and Tail	1.4 cm, 2.3 cm	B-catenin, progesterone receptor, CD-10	+	Watchful waiting	Alive – no recurrence	4 years

Abbreviations: ND, not described; PD, pancreaticoduodenectomy.

currence after surgical resection. Desmoid tumors are nearly 1,000 times more common in patients with FAP compared to the general population and occur in approximately 10% of those affected by FAP [5–7]. Our patient had large multiple desmoid tumors of the abdominal wall, chest wall, and paraspinal soft tissues, that exerted extrinsic compression on nearby structures.

Pancreatic tumors are rare extracolonic manifestations, developing mostly as ductal adenocarcinomas. The lifetime risk of pancreatic carcinoma in patients with FAP is low, at about 2%, but the risk is 4 times higher than that of the general population [8]. However, it rarely co-occurs with solid pancreatic pseudopapillary neoplasm (SPN). SPN is a low-grade malignant neoplasm, predominantly occurring in young women, composed of poorly organized, monomorphic epithelial cells forming solid and pseudopapillary structures and frequently undergoing hemorrhagic-cystic degeneration [9]. It constitutes about 0.17%-2.7% of pancreatic tumors and can be cured by complete resection [2,10].

The connection between the unusual pair is likely driven by a genetic predisposition. In the majority of solid pseudopapillary neoplasm (SPN) cases, missense mutations in exon 3 of the β-catenin gene (CTNNB1) have been identified, leading to elevated β-catenin levels in such patients [10]. β-catenin is an important component of the Wnt/β-catenin signaling that controls cell proliferation, differentiation, and development [11]. Mutations in the APC gene, in cases of FAP, activate the Wnt/β-catenin signaling pathway, resulting in increased levels of cytoplasmic β-catenin [12]. Consequently, individuals with FAP may develop SPN due to alterations in the cytoplasmic β-catenin levels.

According to the literature, 7 cases of SPN occurring together with FAP have been reported. A summary of all reported cases, including our case, is provided in Table 1. Interestingly, 6 of the 8 reported cases were young females, which is consistent with typical demographic distributions for SPNs. Six of them had positive immunostaining for β-catenin, the possible driver for SPN in FAP patients. Four reported cases had tumors in the head of the pancreas, while 2 were located in the tail. Notably, there has yet to be a documented case involving multiple SPNs with FAP, as observed in our patient, who presented with multifocal SPNs— located in the head and tail of the pancreas.

It is extremely important to report such cases to be able to diagnose rare conditions occurring concurrently. Serial surveillance with CT scans is important in patients with FAP to look for extracolonic manifestations like desmoid tumors or rare co-occurring tumors, like SPN, especially in vulnerable populations i.e., young females, as they have a favorable outcome postresection.

Conclusion

The simultaneous presentation of FAP and SPN in a single patient is exceedingly rare. This case reiterates the critical role of radiological imaging in the detection and management of such complex cases. Coordinated efforts between radiologists and pathologists are essential for achieving an accurate diagnosis and formulating an effective treatment strategy, which may or may not include surgical coordination in cases like this. Regular surveillance and early intervention are paramount in managing patients with FAP to prevent the likely progression of both colonic and extracolonic manifestations.

Author contributions

All authors contributed equally to the writing of this manuscript.

Patient consent

The patient reported in the manuscript signed the informed consent/authorization for participation in research, which includes the permission to use data collected in future research projects such as the presented case details and images used in this manuscript.

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