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Case Report / Приказ болесника

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Familial adenomatous polyposis and colorectal cancer – how sensitive is computed tomography in detecting the underlying disease?

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SUMMARY

Introduction Familial adenomatous polyposis is an autosomal dominant disorder characterized by the presence of 100 or more adenomatous polyps in the mucosal lining of the large intestine with a significant risk of colorectal cancer development.

Case outline This article presents a case report of previously undiagnosed familial adenomatous polyposis in a patient admitted to the surgical emergency department with suspected sigmoid carcinoma. On computed tomography (CT), the findings of the colonic mucosa were inconclusive due to inadequate distension of the bowel lumen and insufficient preparation. Edema of the bowel wall was clearly observed, a CT characteristic of the carcinoma that had formed at the level of the sigmoid colon, while the two other foci of malignant transformation were obscured by a diffuse, uniform thickening of the wall, which was clearly diagnosed as familial adenomyomatous polyposis on subsequent colonoscopy. The patient underwent a total proctocolectomy, after which he continued his oncological treatment.

Conclusion Computed tomography is inadequate for the diagnosis of diffuse polyposis of the colonic mucosa, especially in emergency situations when patients are not prepared for the examination, i.e. without sufficient dilation of the bowel lumen. Since the underlying disease in this patient masked two of the three malignant lesions of the colon, we point out the diagnostic inferiority of the CT examination in the regular emergency settings, to CT and MR colonography and especially to colonoscopy as the gold standard in the detection of colorectal cancer.

Keywords: familial polyposis; colorectal cancer; computed tomography; total proctocolectomy

САЖЕТАК

Увод Породична аденоматозна полипоза је аутозомно доминантно обољење које карактерише присуство 100 или више аденоматозних полипа на слузокожи дебелог црева што представља предиспозицију за развој колоректалног карцинома.

Приказ болесника Овај рад представља приказ случаја претходно недијагностиковане фамилијарне аденоматозне полипозе код болесника примљеног на одељење ургентне хирургије са сумњом на карцином сигмоидног колона. Налаз компјутеризованом томографијом (*CT*) на мукози дебелог црева био је неконклузиван услед недовољне дистензије лумена црева и неадекватне припремљености. Јасно је уочен едем зида, *CT* карактеристика формираног карцинома у нивоу сигмоидног колона док су остала два фокуса малигне алтерације била маскирана дифузним униформним задебљањем зида које је каснијим колоноскопским прегледом јасно дијагностикован као породицна аденомиоматозна полипоза. Болеснику је урађена тотална проктоколектомија након чега је наставио онколошко лечење.

Закључак Компјутеризована томографија је инсуфицијентна у смислу дијагностике дифузне полипозе слузокоже колона нарочито код ургентних стања када пацијенти нису припремљени за преглед, односно без адекватне дистензије лумена цревних вијуга. Како је основно обољење маскирало две од три лезије колоректалног карцинома на дебелом цреву код овог болесника, указујемо и на дијагностичку инериорност *CT* у поређењу са колонографијом помоћу *CT* и *MR*, а нарочито са колоноскопијом као златним стандардом када је у питању откривање колоректалног карцинома.

Кључне речи: породична аденоматозна полипоза; колоректални карцином; компјутеризована томографија; тотална проктоколектомија

INTRODUCTION

Familial adenomatous polyposis (FAP) is characterized by the presence of 100 or more adenomatous polyps in the mucosa of the colon, predisposing it to malignant transformation and contributing significantly to the cumulative risk of developing colorectal carcinoma in patients diagnosed with this disease [1]. FAP can have different inheritance patterns. In most cases, it is an autosomal dominant disease, i.e. one copy of the mutated APC (adenomatous polyposis

coli) tumor suppressor gene is sufficient to cause the disease [2]. This means that the affected persons have a parent who is also affected by the disease. However, if familial adenomatous polyposis is caused by mutations in the MUTYH-associated polyposis (MAP) gene, it is inherited in an autosomal recessive manner [3]. Both copies of the gene must be mutated. In most cases, the parents of a person with an autosomal recessive disease each carry one copy of the mutated gene, but do not show any symptoms of the disease themselves. Therefore, genetic testing is considered the gold standard for FAP diagnosis [4]. Once diagnosed, given the overall risk of colorectal cancer (CRC) from a multifocal adenoma-carcinoma sequence, which often takes years, prophylactic colorectal surgery is the treatment of choice, followed by lifelong endoscopic screening [5].

CASE REPORT

A 42-year-old man presented to the emergency room with left flank pain, nausea, and diarrhea. Anamnestically he reported blood in his stool, and approximated twelve hours had passed since symptom onset. Over the previous year, he had experienced similar episodes of pain and diarrhea, excluding the blood in stool. His medical history was otherwise unremarkable, whereas his family history included his mother's death from colorectal cancer at age 35. Upon physical examination, the patient had left side abdominal sensitivity to palpation.

Abdominal ultrasonography (US) showed a discretely thickened bowel wall of the descending colon, no loop dilation, and hyperechoic left paracolic fat with scarce extraluminal fluid. The US alone was inconclusive. However, that finding, coupled with the leading symptoms at the gastroenterological examination and the complete blood count and laboratory results that yielded a leucocytosis of 12.8×10^9/L, and hemoglobin value of 102 g/L warranted a computerized tomography (CT) exam.

A subsequent CT scan revealed diffuse, irregular thickening of the colonic wall, with a lobulated luminal contour and discrete paracolic stranding of the fat plane (Figure 1). Along the wall of the colon, discrete intraluminal nodulations of the mucosa could be observed, which could not be adequately interpreted due to the inadequate preparation of the colon for examination (Figure 2A).

Distally, in the sigmoid colon, the CT also showed a focal, circular wall thickening, which is incompatible with an inflammatory etiology and most likely corresponds to a malignant change (Figure 2B). The patient was referred and a colonoscopy was performed to gain further insight

into the condition of the suspicious mucosa. It revealed hundreds of polyps throughout the colon with mild inflammation of the mucosa and no mucosal preservation, with the exception of a small segment in the coecum. Most of the polypous lesions were 10 millimeters in size, whereas 10-20 of them were 20-25 millimeters in diameter (Figure 3A). In addition, on examination, an ulcero-infiltrative lesion was noted in the sigmoid colon, approximately 30 centimeters from the anocutaneous line, involving the entire circumference of the colon, extending proximally for approximately 10 centimeters, with relative luminal stenosis, but allowing passage of the endoscope. The lesion bled during the biopsy (Figure 3B).

The histopathologic findings of the biopsied sigmoid mucosa confirmed a malignant change in the sense of an evolved adenocarcinoma, which was a sufficient indication for surgical treatment. Consequently, a total proctocolectomy with ileo-anal anastomosis and bipolar ileostomy was performed. The macroscopic and microscopic specimens of surgical pathology are shown and described below (Figure 4).

The authors confirm that they have read the journal's position on issues involving ethical publication and affirm that this work is consistent with those guidelines. All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Written consent to publish all shown material was obtained from the patient.

DISCUSSION

Familial adenomatous polyposis is characterized by the presence of 100 or more adenomatous polyps in the mucosa of the colon, predisposing it to malignant transformation and contributing significantly to the cumulative risk of developing colorectal carcinoma in patients diagnosed with this disease [1]. FAP can have different inheritance patterns. In most cases, it is an autosomal dominant disease, meaning that one copy of the mutated adenomatous polyposis coli (APC) tumor suppressor gene in each cell is sufficient to cause the disease [2]. This means that affected individuals have a parent who also suffers from the disease. However, if familial adenomatous polyposis is caused by mutations in the MUTYH-associated polyposis (MAP) gene, it is inherited in an autosomal recessive manner [3]. Both copies of the gene must be mutated in each cell. In most cases, the parents of a person with an autosomal recessive disease each

carry one copy of the mutated gene, but do not show any symptoms of the disease themselves. Therefore, genetic testing is considered the gold standard for FAP diagnosis [4].

The family members of the proband are invited for screening before symptoms appear. The National Comprehensive Cancer Network (NCCN) guideline for screening for APC gene mutations recommends testing individuals at the age of 10 years and starting screening endoscopies at the age of 10–15 years [5].

They undergo prophylactic surgery as a preventive measure against cancer. The risk of developing cancer before the age of 20 is very low at only 1% of all FAP patients [6]. It is currently recommended that prophylactic surgery be performed at the age of 20 or 25 [7].

In an emergency situation where no genetic testing had been performed and there is no any information about possible FAP, prior to the onset of symptoms and even when the family history is positive for CRC in young age, it is not surprising that the CT exam did not reveal diffuse mucosal polyposis, probably due to inadequate expansion of the bowel lumen and the presence of residual contents. However, as the underlying genetic disease successfully masked two of the three CRC lesions, we want to emphasis that CT scan is inferior to MRI and CT colonography in terms of sensitivity when it comes to detecting CRC [8].

Conflict of interest: None declared.

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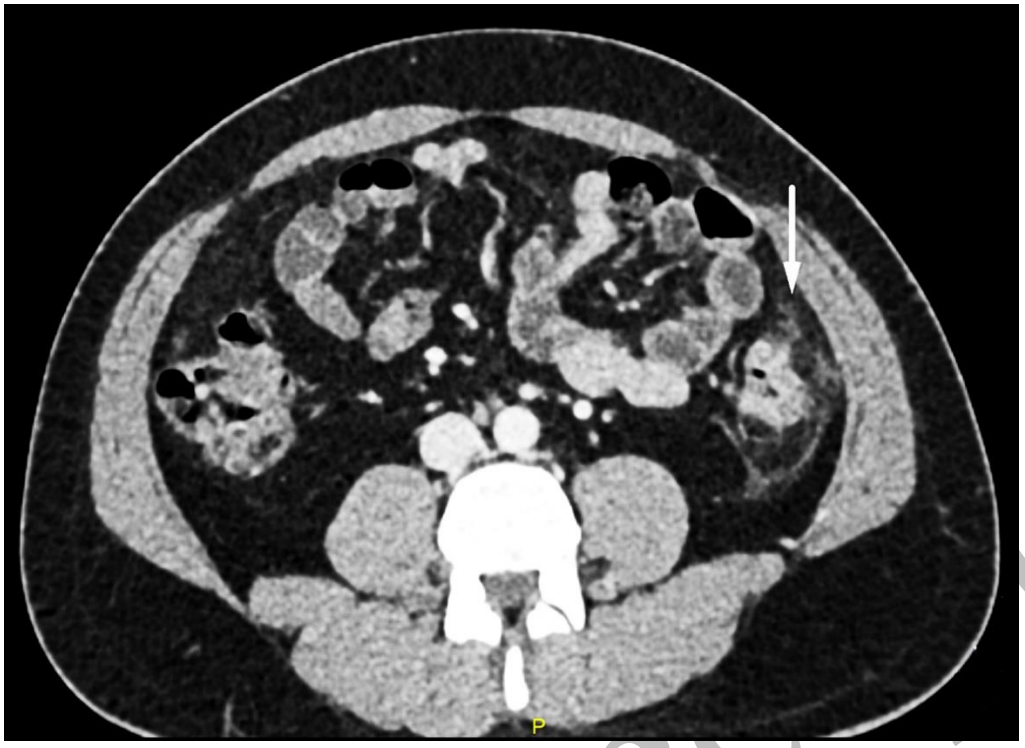

Figure 1. A contrast-enhanced axial CT scan showing bowel wall thickening in the descending colon (white arrow) with a "fat-stranding" pattern regionally

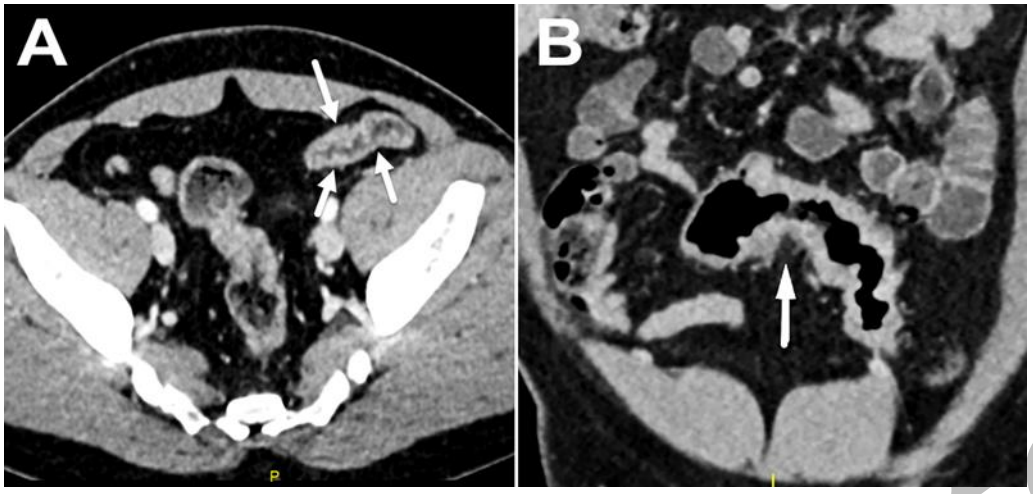

Figure 2. Contrast-enhanced axial (A) and coronal (B) CT scans showing the segmental (white arrows) thickening of the sigmoid colon wall highly suspicious of a neo-infiltrative process, but without clear visualization of multiple polyps on the mucosa

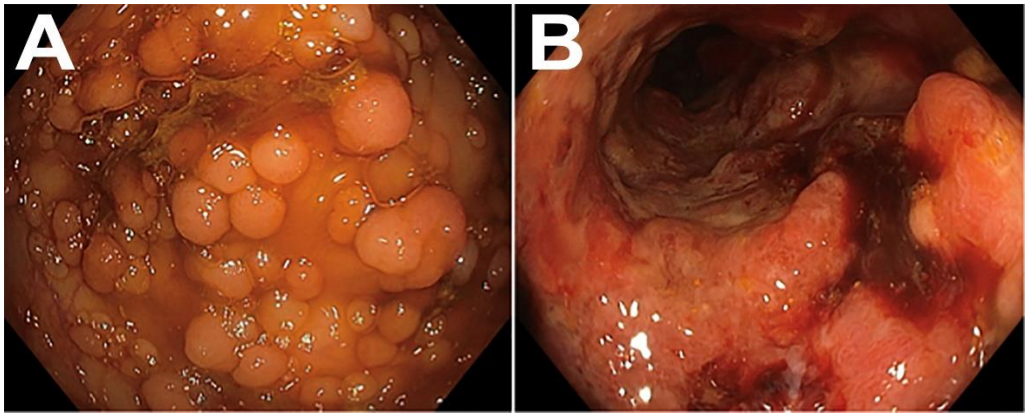

Figure 3. Colonoscopy findings in the transverse (A) and the sigmoid colon (B), showing a multitude of subcentimetric polyps with no healthy remaining mucosa and the previously described ulcero-infiltrative lesion, respectively

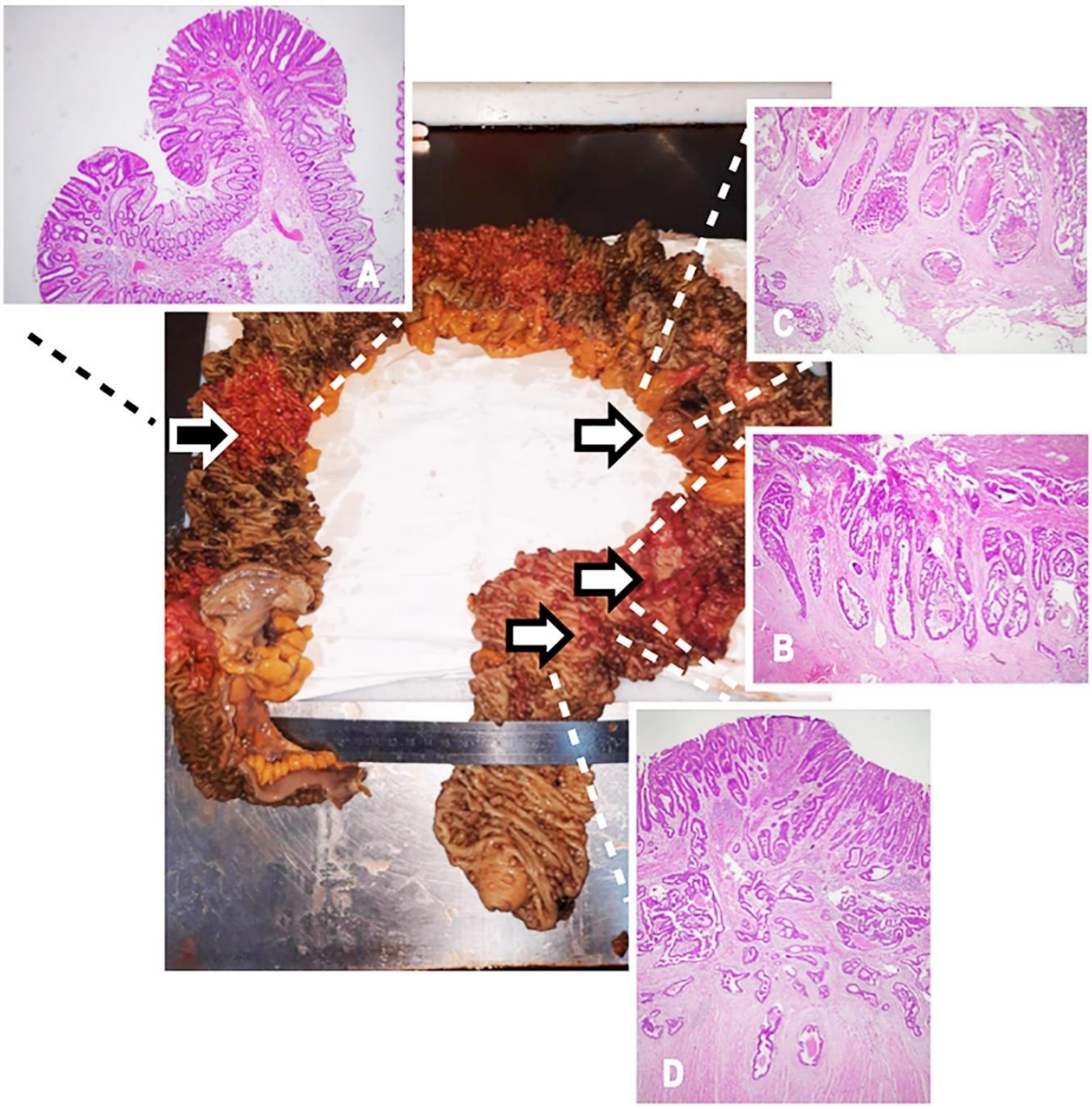

Figure 4. The intestinal resection specimen included a 112-cm-long colon, an 18-cm-long portion of the terminal ileum, and a 55-mm-long appendix; innumerable polyps were present throughout the entirety of the colorectal segment, from the Bauhin's valve all the way to 2–3 mm proximal to the lower resection line, with a dispersed distribution; most of them were sessile and semi-sessile, 2–10 mm in diameter, while a handful were pedunculated, polypoid, exceeding that size (black arrow); a photomicrograph of one such polyp is shown above (A); macroscopically, the resection specimen showed three well-demarcated tumors, two of which in the sigmoid and one in the descending colon (white arrows); the CT-suspected lesion was ulcero-vegetative, affecting the whole circumference of the sigmoid, with the infiltration depth of 10 mm and with a mixed tubular/tubulo-cribriform organization and expansive-infiltrative growth with signs of extramural invasion, microscopically (B); another tumor, smaller in size, was located proximally in the descending colon, also presenting as an ulcerative vegetative

lesion, which demonstrated an infiltration depth of 10 mm (C); distal to the former the ulcerative-vegetative tumor of the sigmoid colon, a larger sessile polyp was observed within the same region and biopsied; the polyp showed suspicious infiltration into the submucosa and muscle layer on macroscopic cross-sections, a finding that was subsequently confirmed through histopathological examination as CRC (D); microscopically, both tumors (C and D) were mixed tubular/tubulo-cribriform adenocarcinomas, the first of which showed signs of extramural invasion (C), whereas the second one was intramural growth only (D)

