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CASE REPORT / ПРИКАЗ БОЛЕСНИКА

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

Sofija Jovanović[1], Tijana Tomić[1], Nemanja Bidžić[2,3], Miloš Raspopović[4], Dragan Vasin[1,5]

[1]University Clinical Centre of Serbia, Center for Radiology and Magnetic Resonance Imaging, Belgrade, Serbia;
[2]University Clinical Centre of Serbia, Clinic for Digestive Surgery, Department for HPB Surgery, Belgrade, Serbia;
[3]University of Belgrade, Faculty of Medicine, Department for Surgery, Belgrade, Serbia;
[4]University of Clinical Center of Serbia, Clinic for Emergency Surgery, Belgrade, Serbia;
[5]University of Belgrade, Department for Radiology, Faculty of Medicine, Belgrade, Serbia

SUMMARY

Introduction Familial adenomatous polyposis (FAP) 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 FAP 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 FAP 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, compared 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

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 cancer (CRC) 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 adenomatous polyposis coli (APC) tumor suppressor gene is sufficient to cause the disease [2]. This means that the affected individuals have a parent who is also affected by the disease. However, if FAP 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 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 estimated that 12 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 CRC at age 35. Upon physical examination, the patient had left-sided abdominal tenderness on 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.

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Correspondence to:
Dragan VASIN
Pasterova 2
11000 Belgrade, Serbia
draganvasin@gmail.com

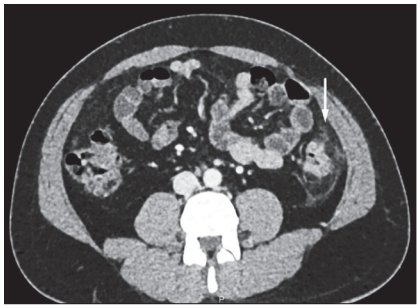

Figure 1. A contrast-enhanced axial CT scan showing bowel wall thickening in the descending colon (white arrow) with a "fat-stranding" pattern regionally

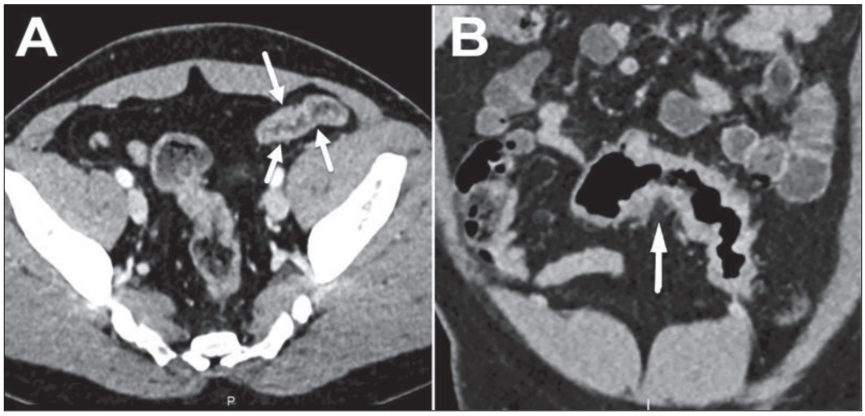

Figure 2. Contrast-enhanced axial (A) and coronal (B) CT scans showing the segmental (white arrows) thickening of the sigmoid colon wall highly suspicious of a neo-infiltrative process, but without clear visualization of multiple polyps on the mucosa

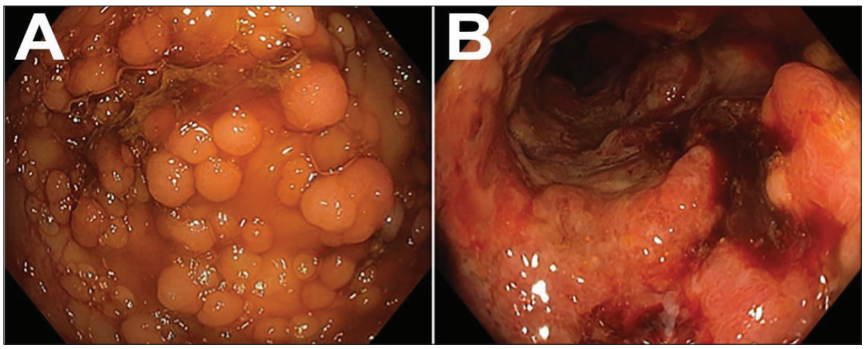

Figure 3. Colonoscopy findings in the transverse (A) and the sigmoid colon (B), showing a multitude of subcentimetric polyps with no healthy remaining mucosa and the previously described ulcero-infiltrative lesion, respectively

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 leukocytosis measuring 12.8×10^9/L and hemoglobin 102 g/L warranted a computed 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, except for a small segment in the cecum. Most of the polypus lesions were 10 mm in size, whereas 10–20 of them were 20–25 mm in diameter (Figure 3A). In addition, on examination, an ulcero-infiltrative lesion was noted in the sigmoid colon, approximately 30 cm from the anocutaneous line, involving the entire circumference of the colon, extending proximally for approximately 10 cm, 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 transformation consistent with an advanced 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

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 CRC 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 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 FAP is caused by mutations in the 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

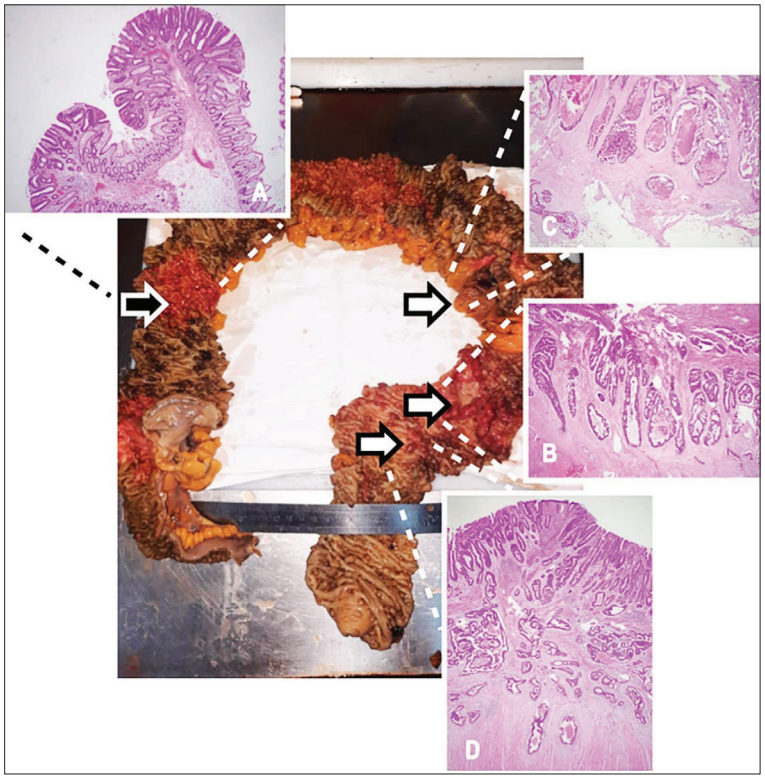

Figure 4. The intestinal resection specimen included a 112-cm-long colon, an 18-cm-long portion of the terminal ileum, and a 55-mm-long appendix; innumerable polyps were present throughout the entirety of the colorectal segment, from the Bauhin's valve all the way to 2–3 mm proximal to the lower resection 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)

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 guidelinec 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 years 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 has been performed and where there is no information about possible FAP prior to the onset of symptoms, and even when the family history is positive for CRC at a 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 emphasize that a 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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Породична аденоматозна полипоза и колоректални карцином – колико је компјутеризована томографија осетљива метода за откривање основне болести?

Софија Јовановић[1], Тијана Томић[1], Немања Бицић[2,3], Милош Распоповић[4], Драган Васин[1,5]

[1]Универзитетски клинички центар Србије, Центар за радиологију и магнетну резонанцу, Београд, Србија;
[2]Универзитетски клинички центар Србије, Клиника за дигестивну хирургију, Одељење за хепатобилиопанкреатичну хирургију, Београд, Србија;
[3]Универзитет у Београду, Медицински факултет, Катедра за хирургију, Београд, Србија;
[4]Универзитетски клинички центар Србије, Клиника за ургентну хирургију, Београд, Србија;
[5]Универзитет у Београду, Медицински факултет, Катедра за радиологију, Београд, Србија

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 породичне аденоматозне полипозе код болесника примљеног на одељење ургентне хирургије са сумњом на карцином сигмоидног колона. Налаз добијен компјутеризованом томографијом на мукози дебелог црева био је неконклузиван услед недовољне дистензије лумена црева и неадекватне припреме. Јасно је уочен едем зида, налаз на компјутеризованој томографији карактеристичан за формирани карцином у пределу сигмоидног колона, док су остала два фокуса малигне алтерације била маскирана дифузним, униформним задебљањем зида, које је каснијим колоноскопским прегледом јасно дијагностиковано као породична аденоматозна полипоза. Болеснику је урађена тотална проктоколектомија, након чега је наставио онколошко лечење.

Закључак Компјутеризована томографија је недовољно осетљива метода за дијагностику дифузне полипозе слузокоже колона, нарочито код ургентних стања када болесници нису припремљени за преглед, односно када није обезбеђена довољна дистензија лумена цревних вијуга. Како је основно обољење маскирало две од три лезије колоректалног карцинома на дебелом цреву код овог болесника, указујемо и на дијагностичку инфериорност компјутеризоване томографије у поређењу са колонографијом помоћу компјутеризоване томографије и магнетне резонанце, 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