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Case report

Unusual presentation of small bowel obstruction in Cowden syndrome: A case report and literature review

Natalia Dardzińska, Emil Obrębski, Dominika Lorek, Paulina Jaskulska, Natalia Krzysztofek, Kamil Jończyk, Bartosz Molasy

Collegium Medicum, Jan Kochowski University in Kielce, Poland

Address for correspondence:
Natalia Dardzińska
Collegium Medicum
Uniwersytet Jana Kochanowskiego w Kielcach
Aleja IX Wieków Kielc 19A
25-317 Kielce
e-mail: nataliad0609@icloud.com

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ABSTRACT

Intestinal intussusception in adults is a rare disorder, occurring in about 5% of cases. In the pediatric population, it is most often idiopathic in origin, while in adults it is associated with pathologies within the intestinal wall. The purpose of this study is to emphasize the need for expanded diagnosis in adult patients with symptoms of gastrointestinal obstruction due to intestinal intussusception. The medical records of a 22-year-old female patient hospitalized in the Department of General Surgery of St. Alexander Hospital in Kielce were analyzed. The patient presented to the hospital with complaints of abdominal pain accompanied by vomiting. She had a history of treatment for Hashimoto's disease, iron deficiency anemia and gastritis. After initial diagnosis, the patient was transferred to the department of general surgery, where conservative treatment was implemented and gastroscopy was performed. During hospitalization, the patient was consulted in internal medicine due to uncompensated hypothyroidism. Due to lack of improvement, imaging studies were ordered, which revealed a dilated loop of small intestine with suspected intussusception. The patient was qualified for surgery, during which an intussusception of the jejunum was found. Due to the inability to drain the intussusception, a segmental small bowel resection was performed. Histopathological examination revealed a hamartomatous polyp as the cause of the intussusception. Given the overall clinical picture, the patient was referred to the genetic counseling center, where Cowden syndrome was confirmed. Intestinal intussusception, although rare, should be considered as a cause of gastrointestinal obstruction in adults. Identification of the pathological changes that are its cause is crucial to the overall diagnostic and therapeutic process.

KEYWORDS

Cowden syndrome, hamartomatous polyp, intussusception

INTRODUCTION

Intussusception, a form of intestinal obstruction, is often described as the intussusception of one segment of intestine into an adjacent segment. The vast majority of cases of intestinal intussusception are treated by pediatric surgeons, as the condition is more common in children than in adults. It is estimated that about 5% of all intussusception cases occur in adults [1]. Considering all cases of intestinal obstruction in adults, only about 1% are due to intussusception. In adults, these cases are usually associated with pathological conditions, while in children they are usually idiopathic [2,3]. The most common underlying pathology in adult intussusception is a benign tumor (about 37%). Most of the lesions are in the intestinal location [4]. Hamartomatous polyps may be associated with Cowden's syndrome and may pose a risk of intussusception, which can lead to intestinal obstruction [5,6]. Cowden syndrome (CS) is a rare genetic disorder caused by mutations in the phosphatase and tensin homolog gene (PTEN) that most commonly affects young people in

the second and third decades of life [7]. The incidence is about 1 in 200,000, slightly more common in women [8]. Among other things, the disease is characterized by multiple hamartomatous lesions occurring in various organs [9,10]. It is most commonly manifested by cutaneous and mucosal lesions, which include acral keratosis or oral papillomatosis [11,12]. The syndrome leads to an increased risk of malignancies including thyroid and colorectal cancer (CC) [13]. The PTEN gene plays a key role in gastrointestinal (GI) and cancerous processes, including hamartomatous polyposis syndromes (HPS), which are genetic conditions characterized by polyps in the GI tract [14]. Individuals with CC are often found to have preexisting colorectal polyposis. Most individuals with at least one mutant allele of the PTEN gene have polyps in the large intestine in 90%-95% of cases [11]. Polyps also occur in the small intestine, but the risk of tumorigenesis is not specified in the literature [15].

The medical data of a 22-year-old female patient were retrospectively reviewed, including clinical presentation, laboratory and imaging studies, and surgical findings. Abdominal computed tomography (CT) scans, gastroscopy, and histopathological examination of resected intestinal specimens were analyzed. The diagnostic and therapeutic approach was consistent with current clinical guidelines for suspected small bowel obstruction and intussusception. Informed consent for publication was obtained from the patient.

CASE REPORT

A 22- year-old female patient presented to the Emergency Department with complaints of abdominal pain and associated vomiting of gastric contents. Physical examination revealed tenderness to palpation in the mid-epigastric and left mid-abdomen regions, accompanied by increased tension of the abdominal wall. Peritoneal symptoms were not found. In addition, the abdomen was slightly bloated, peristalsis was preserved, and there were no abnormalities on per rectum examination. The patient has been treated for anemia and hypothyroidism since she was 5 years old. In addition, she is under gynecological care due to heavy menstruation. As a result, the patient is taking Euthyrox, iron and ad hoc Cycloamine. On admission, vital signs were normal. On the NRS scale, the patient reported a pain score of 7/10. In laboratory tests, a case of severe anemia with a hemoglobin level of just 4.2 g/dLwas observed. The mean corpuscular volume (MCV) was notably low, recorded at 66.56 fL on September 20, 2024, and it fluctuated between 76.28 and 80.6 fL during the hospital stay. Additionally, serum iron levels were low, showing 24 µg/dL on September 23, 2024, and dropping to 18 µg/dL the following day (Table I). The patient was transfused with 3 units of red blood cell concentrate (RBC) and conservative treatment was implemented. Given her medical history, the patient was consulted gynecologically. Due to nonspecific complaints of abdominal pain, a computed tomography (CT) scan of the abdomen and pelvis without contrast was performed. Based on the examination, intussusception of the bowel was

suspected. The patient was admitted to the department of general surgery, where conservative treatment was continued; the patient was given gastrografin. An internal medicine consultation was required due to uncompensated hypothyroidism. Gastroscopy was also performed, which revealed superficial gastritis. During hospitalization, the patient developed extrapyramidal symptoms after administration of metoclopramide, which necessitated a head CT scan. On the second day after admission, a follow-up CT scan of the abdomen and pelvis was performed, which showed free abdominal fluid and a dilated loop of intestine, again raising the suspicion of intestinal intussusception. Considering the results of the follow-up abdominal CT scan and the lack of improvement after conservative treatment, the patient was qualified for urgent surgery. Intraoperatively, distended loops of bowel and a significant amount of fluid in the abdominal cavity were found. Due to the inability to perform the procedure safely by laparoscopy, the decision was made to convert to laparotomy. After palpation inspection of the abdominal cavity, an intussusception of the jejunum about 5 cm from the ligament of Treitz was found.

Table I. Laboratory findings during hospitalization

Date	MCV (fL)	Serum Iron (µg/dL)
2024/09/20	66.56	–
2024/09/21	76.28–76.55	–
2024/09/22	77.43	–
2024/09/23	78.09	24
2024/09/24	78.9	18
2024/09/26	80.6	–
2024/09/28	80.25	–
2024/09/29	79.64	–
2024/09/30	79.77	–

Due to the inability to drain the intussusception, the decision was made to perform a segmental resection of the jejunum. During the preparation of the bowel segment, spontaneous drainage of the intussusception occurred, thus making visible the microperforation of the jejunum. After resection of the bowel segment, an end-to-side anastomosis was performed, and the patency and blood supply of the anastomosis were checked. A Redon drain was deposited into the smaller pelvis, and the wound was sutured in layers. The bowel fragment was submitted for histopathological examination with a suspected proliferative lesion of the jejunum. The postoperative period went without complications. An oral diet was gradually expanded, which the patient tolerated well. She was discharged home on the eighth postoperative day. In view of the overall clinical picture, the patient was referred to the genetic counseling center, where Cowden syndrome was confirmed.

DISCUSSION

In the United States, the incidence of small bowel obstruction (SBO) is estimated to be approximately 350,000 cases per year. The most common causes are adhesions (65%), hernias (10%), cancer (5%), and Crohn's disease (5%) [16].

Invagination of the intestine (intussusception) occurs mainly in the pediatric population, where it is most often not accompanied by a discernible structural cause. In adults, this condition is rare – accounting for about 5% or 1 % of all intussusception cases – and is characterized by a nonspecific clinical presentation, which significantly complicates accurate diagnosis [1,2,3,17]. Adult intussusception is often associated with structural lesions, with tumors or polyps accounting for approximately 35% of cases [18]. A meta-analysis of 1,229 patients showed malignancy in 32.9%, benign lesions in 37.4%, and idiopathic etiology in 15.1% of cases [4]. Structural causes were identified in 85.7% of cases, including 35.3% benign and 64.7% malignant tumors [17]. Lead points may include fibroid polyps or submucosal lipomas [19,20]. A 22-year-old patient was admitted with intestinal obstruction resulting from intussusception caused by a juvenile hamartomatous polyp. Genetic testing confirmed a PTEN mutation. Up to 95% of adults with PTEN mutations develop polyps, which can be morphologically diverse, including adenomas, hamartomas, juvenile, lipomatous, leiomyomatous, lymphoid, and hyperplastic polyps [21]. According to the NCCN diagnostic criteria, Cowden syndrome/PTEN hamartoma syndrome (PHTS) is diagnosed based on the presence of major features (e.g., breast cancer, endometrial cancer, follicular thyroid cancer, macrocephaly, adult Lhermitte-Duclos disease, gastrointestinal hamartomas, or multiple characteristic mucocutaneous lesions) and secondary features (e.g., colorectal cancer, renal cell carcinoma, autism spectrum disorder, lipomas, or vascular malformations). A clinical diagnosis is established when an individual meets at least 3 major criteria (including at least one of the following: macrocephaly, Lhermitte-Duclos disease, or gastrointestinal hamartomas) or 2 major and at least 3 secondary criteria. In families with a confirmed PTEN mutation or a known clinical diagnosis of PHTS, it is sufficient to meet 2 major criteria or 1 major and 2 minor criteria or at least 3 minor criteria [10].

It is worth noting that intussusception in adults is rare, but when it does occur, it is often accompanied by a cancerous change, as was the case in our situation. It should also be remembered that in young patients – like our case– the symptoms of CS may be incomplete, which requires special diagnostic vigilance.

Computed tomography (CT) is the first-line test in the diagnosis of intestinal intussusception and allows both the identification of the cause and the planning of appropriate treatment [17]. X-ray of the abdominal cavity can be helpful as an initial examination in patients with suspected intestinal obstruction. Magnetic resonance imaging (MRI) offers greater accuracy in assessing blockages, but due to the cost and time required, it remains a supplementary method [22]. For patients in stable

general condition with abdominal surgery history, conservative treatment should be considered first. Oral administration of a hypertonic, water-soluble contrast agent may have a therapeutic effect and accelerate the resolution of the obstruction. Research has shown that such treatment reduces the need for surgical intervention and shortens the duration of hospitalization [22,23]. In case of a worsening clinical condition, urgent surgical intervention is necessary [22]. The choice between laparoscopy and laparotomy depends on the anatomical conditions and the patient's general condition. In case of small intestine obstruction, the laparoscopic technique may be difficult due to the distension of the intestinal loops and the thinning of their walls, which increases the risk of perforation [24]. In clinical practice, a hybrid approach is often used, starting laparoscopically and converting to laparotomy if necessary [25] as was the case in the one being discussed. Comparing conservative treatment with surgical treatment of intestinal obstruction in patients who underwent surgical treatment, a lower recurrence rate is noted; however, the length of hospitalization in this group was longer compared to patients treated conservatively [26]. Laparoscopy, compared to laparotomy, is usually associated with a shorter procedure and a shorter hospital stay [27]. Limitations of this report include the absence of genetic testing in family members, which could provide valuable information about inheritance patterns. Additionally, the short postoperative follow-up period does not allow for assessment of long-term outcomes or recurrence. Finally, this case report's findings cannot be generalized due to its single-patient nature.

CONCLUSIONS

Intestinal intussusception in adults, although rare, can be the first clinical manifestation of Cowden syndrome. The described case emphasizes the importance of considering genetic syndromes in young adults presenting with nonspecific gastrointestinal symptoms, especially when coexisting with endocrine and hematologic disorders. Surgical intervention remains the mainstay of treatment in cases of bowel obstruction caused by intussusception, while genetic counseling and lifelong surveillance for associated malignancies are essential in confirmed cases of Cowden syndrome.

Authors' contribution

Study design – N. Dardzińska, B. Molasy,

Data collection – N. Dardzińska, E. Obrębski

Data interpretation – N. Dardzińska, B. Molasy

Manuscript preparation – N. Dardzińska, E. Obrębski, D. Lorek, P. Jaskulska, N. Krzysztofek, K. Jończyk

Literature research – N. Dardzińska, E. Obrębski

Final approval of the version to be published – B. Molasy

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