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E-mail: info@galenos.com.tr/yayin@galenos.com.tr

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CASE REPORTS

CASE REPORT

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J Cau Med Sci 2025;3(1):1-3

Perforated Non-Meckel Ileal Diverticulum Mimicking Neoplastic Lesion: A Case Report

Shukufa Musayeva[1], Hazi Aslanov[2], Ilaha Karimova[3]

[1]Azerbaijan Medical University, Department of Surgical Diseases, Baku, Azerbaijan
[2]Scientific Surgical Center named after Academician M. Topchubashov, Department of General Surgery, Baku, Azerbaijan
[3]Azerbaijan Medical University, Department of Pathology, Baku, Azerbaijan

ABSTRACT

Non-Meckel small bowel diverticulosis is a rare condition often asymptomatic but capable of causing complications such as hemorrhage, perforation, and obstruction. It primarily affects elderly men, with the duodenum being the most common site. The pathogenesis is linked to myenteric plexus dysfunction, leading to increased intraluminal pressure and wall herniation. This case study presents a 58-year-old male admitted with acute abdominal symptoms. Imaging revealed free peritoneal fluid and intraperitoneal gas, prompting emergency laparotomy. Early diagnosis and appropriate treatment are crucial for preventing severe outcomes. Histopathological examination is necessary to rule out malignancy, particularly in emergency cases. Small bowel diverticulosis should be considered in differential diagnoses of acute abdomen.

Keywords: Ileum, Non-Meckel diverticulum, Complication, Acute abdomen, Surgery

INTRODUCTION

A diverticulum is an abnormal sac or pouch that protrudes from the wall of a hollow organ [1]. In the gastrointestinal tract, these pouches commonly develop in the esophagus, small intestine, or large intestine, with the colon being the most frequently affected site. The formation of diverticula typically occurs in weak areas of the bowel wall where blood vessels penetrate [2].

Most diverticula occur in the sigmoid colon; however, they can also be found in the descending colon (40%) and, less frequently, in other parts of the colon (5-10%). In contrast, non-Meckel small bowel diverticular disease is rare, with a prevalence of 0.01-2.3%. Autopsy studies have detected small bowel diverticula in 0.3-1.3% of cases, while small intestine contrast studies have identified them in 0.5-1.9% of the population. This condition primarily affects elderly men, with the duodenum being the most common site of occurrence, followed by the jejunum and ileum [3,4].

Non-Meckel small bowel diverticulosis is predominantly asymptomatic but may present with diarrhea, malabsorption, or chronic abdominal pain. Acute complications, including hemorrhage, perforation, fistulas, diverticulitis, and intestinal obstruction, occur in 10-15% of cases [4].

This study presents a case of ileal non-Meckel diverticulum with hemorrhage and perforation in a 58-year-old male. The clinical, surgical, and pathological features of the patient are discussed.

CASE REPORT

A 58-year-old male patient was admitted to the hospital with symptoms of acute abdomen. Ultrasonography revealed free fluid in the peritoneal cavity, and an X-ray examination showed intraperitoneal gas. The patient underwent emergency laparotomy. During the procedure, an exophytic mass with bleeding and necrotic changes, measuring 6×7×10 cm, was observed in the ileal wall. A significant amount of liquid blood and blood clots was aspirated from the peritoneal cavity. The affected ileal segment and a portion of the greater omentum were resected.

Gross examination of the specimen revealed that the ileal mass was a large perforated diverticulum (Figures 1 and 2). Histopathological analysis showed acute non-specific inflammation and gangrenous necrosis in the diverticular wall, with no evidence of ectopic tissue. The patient was discharged on the 10th postoperative day without complications. No

Address for Correspondence: Shukufa Musayeva MD, Azerbaijan Medical University, Department of Surgical Diseases, Baku, Azerbaijan
E-mail: shukufamusa@gmail.com **ORCID ID:** 0000-0002-1206-8323
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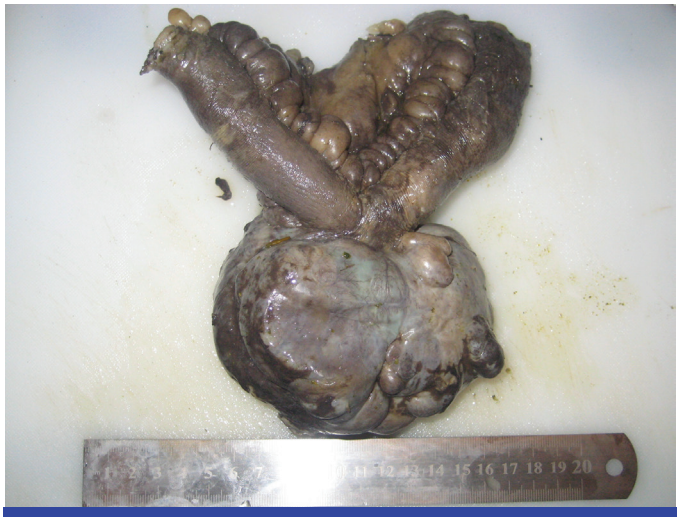

Figure 1. Macroscopic view of resected small bowel and perforated diverticulum

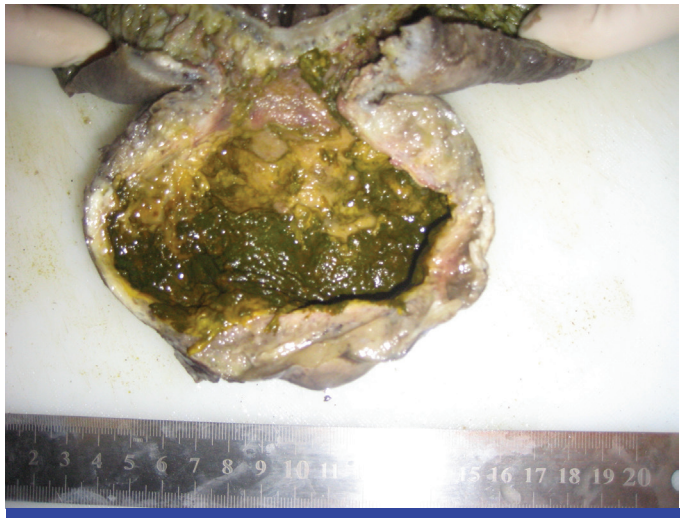

Figure 2. The diverticular cavity containing stool pieces and its relationship with the lumen of the small intestine

adverse events were observed during the 12-month follow-up period.

DISCUSSION

Diverticular disease is often referred to as a "Western disease" due to its higher prevalence in European and North American populations, whereas significantly lower rates are observed in African and Asian countries. This discrepancy is believed to be linked to a combination of genetic factors and dietary habits, particularly the lower fiber intake typical of Western diets [5]. It is a common condition, especially among the elderly, affecting 10% of individuals over 40 and up to 50% of those over 60. It is rarely seen in individuals under 40, with only 2-5% of cases occurring in this age group. Both genders are equally affected, though men tend to develop diverticular disease and diverticulitis at a younger age (under 50) [3,4].

Individuals between the ages of 50 and 70 who consume a high-fiber diet (at least 25 grams per day) are 40% less likely to be hospitalized for complications of diverticular disease, compared to those with lower fiber intake. This underscores the crucial role of fiber in the prevention and management of diverticular conditions [6].

Most small bowel diverticula are false diverticula, lacking a muscularis layer, unlike true diverticula such as Meckel's. The pathogenesis of small bowel diverticulosis is thought to be related to motor dysfunction of the myenteric plexus, leading to disordered bowel contractions, increased intraluminal pressure, and subsequent herniation at sites where blood vessels and nerves penetrate the bowel wall. The jejunum is more frequently affected due to the diameter of its penetrating arteries [4].

Ileal diverticulosis, although less common than duodenal and jejunal diverticulosis, is more prone to complications such as perforation. Up to 19% of patients with ileal diverticulosis experience severe complications, including perforation, often necessitating surgical intervention. Although many cases remain asymptomatic, recurrent symptoms can significantly impact the patient's quality of life [7].

Imaging techniques such as computed tomography (CT), capsule endoscopy, and small bowel follow-through examinations are essential for diagnosing small bowel diverticulosis, particularly in asymptomatic or incidentally discovered cases. In acute presentations, CT scans can reveal bowel wall thickening, mesenteric edema, extraluminal gas, and fluid collection, which are indicative of complications such as perforation. Capsule endoscopy is valuable for diagnosing non-acute cases but less useful in emergency scenarios. Hemorrhagic lesions can be identified using a technetium-99m bleeding scan or arteriography, which detect bleeding rates of 0.1-0.5 mL/min and 0.5-1 mL/min, respectively. Preoperative imaging plays a critical role in surgical planning, particularly in cases of severe complications [6]. In our case, the presence of signs of perforation on ultrasound and X-ray examinations indicated that the patient's condition required urgent surgical intervention. Therefore, other imaging methods were not used.

Although rare, complications such as perforation or hemorrhage require prompt surgical intervention. The coexistence of diverticulosis and small bowel volvulus is an uncommon but serious condition that poses diagnostic challenges. Surgical resection of the affected bowel segment, often with primary anastomosis, is the definitive treatment in such cases. Early diagnosis and careful monitoring are essential to prevent severe outcomes, particularly in elderly patients [7]. Patients with localized abscesses (<3 cm), stable hemodynamics, no signs of peritonitis, and a good response to intravenous antibiotics may be

managed conservatively. Abscesses larger than 3 cm often require percutaneous drainage. Endoscopic therapy is the preferred approach for managing diverticular bleeding when feasible; however, recurrence rates of up to 20% have been reported, necessitating alternative treatments such as angiography or surgery in more severe cases [8]. While surgery is more invasive, it offers definitive treatment and prevents recurrence. Non-Meckel diverticulum of the small bowel is a rare entity that can be complicated by bleeding and perforation. Clinically or intraoperatively, it may mimic a neoplastic mass. Therefore, it should be considered by abdominal surgeons in the differential diagnosis in such cases. Histopathological examination is crucial for excluding malignancy, particularly in patients undergoing emergency surgery.

Ethics

Informed Consent: Written informed consent was obtained from the patient for publication of this case report and the accompanying images.

Footnotes

Authorship Contributions

Surgical and Medical Practices: S.M., H.A., Concept: S.M., H.A., Design: S.M., I.K., Data Collection or Processing: S.M., I.K., Analysis or Interpretation: S.M., H.A., I.K., Literature Search: S.M., Writing: S.M.

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CASE REPORT

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A Rare and Aggressive Abdominopelvic Tumor: A Case of Desmoplastic Small Round Cell Tumor in a Young Male

Yağmur Sena Tosun, Aytan Babazade, Emine Sena Sözen, Betül Erişmiş, Enes Seyda Şahiner

Ankara Bilkent City Hospital, Department of Internal Medicine, Ankara, Turkey

ABSTRACT

Desmoplastic small round cell tumor (DSRCT) is a rare and aggressive mesenchymal tumor primarily affecting young adults, commonly originating in the abdomen and pelvic region. Here, we present the case of a 21-year-old male referred to our facility due to abnormal kidney function tests and a detected abdominal mass. The patient's laboratory results did not reveal any findings suggestive of a specific disease. The patient had experienced back pain for a month, which was managed initially with analgesics. Upon admission, hypertension was noted, and investigations revealed elevated urea, creatinine, uric acid, and lactate dehydrogenase levels. Abdominal ultrasound disclosed a hypoechoic lesion in the bladder pelvis, further confirmed by computed tomography (CT) scans, which showed lung and abdominal metastases. Positron emission tomography/CT scan highlighted hypermetabolic lymph nodes and skeletal lesions. Biopsy confirmed DSRCT with a Ki-67 proliferation index of 15-20%. The patient commenced VAC-IE chemotherapy. Key aspects of interest include the young age, the rapid metastatic spread, and the unexpected diagnosis. This case underscores the diagnostic challenges and aggressive nature of DSRCT, necessitating comprehensive management strategies. Our purpose is to describe the challenges and experiences in the diagnosis of DSRCT.

Keywords: Abdominopelvic tumor, Case report, Desmoplastic small round cell tumor, Young male

INTRODUCTION

Desmoplastic small round cell tumor (DSRCT) is an exceptionally rare and aggressive mesenchymal malignancy that primarily affects adolescents and young adults with a predilection for males [1,2]. The incidence of DSRCT is exceedingly low, with fewer than 500 cases reported in the medical literature since its initial description in 1989, highlighting its status as an orphan disease that poses significant diagnostic and therapeutic challenges [3]. The clinical significance of DSRCT lies in its aggressive behavior, propensity for widespread metastasis, and poor prognosis, necessitating a high index of suspicion and a multidisciplinary approach to management. Due to its insidious onset and non-specific symptomatology, such as abdominal pain, distension, or incidentally discovered palpable masses, DSRCT often poses a diagnostic challenge, frequently resulting in delayed diagnosis, which can significantly impact treatment outcomes and overall survival.

The genetic hallmark of DSRCT is the reciprocal translocation t(11;22)(p13;q12), which results in the fusion of the *EWSR1* gene on chromosome 22 with the *WT1* gene on chromosome 11 [4]. This translocation leads to the formation of the EWSR1-WT1 fusion protein, a chimeric transcription factor that dysregulates the expression of target genes involved in cellular proliferation, differentiation, and apoptosis, thereby driving the pathogenesis of DSRCT [4]. While the EWSR1-WT1 translocation is highly specific for DSRCT, its detection through molecular diagnostic techniques such as fluorescence *in situ* hybridization or reverse transcription polymerase chain reaction is crucial for confirming the diagnosis, particularly in cases with ambiguous histopathological features [4]. It is critical to differentiate DSRCT from other small round blue cell tumors.

CASE REPORT

A 21-year-old male was referred to our facility due to abnormalities in kidney function tests and an incidental finding of an abdominopelvic mass on ultrasound. The patient reported experiencing progressive lower back pain over the past month, which was initially managed with over-

Address for Correspondence: Yağmur Sena Tosun MD, Ankara Bilkent City Hospital, Department of Internal Medicine, Ankara, Turkey
E-mail: ygmrsenat@gmail.com **ORCID ID:** 0009-0008-2892-5319
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the-counter analgesics. Subsequently, he developed nausea and vomiting, prompting further evaluation. Laboratory investigations revealed elevated creatinine, urea, uric acid, and lactate dehydrogenase levels.

On admission, the patient's blood pressure was elevated, requiring antihypertensive treatment. Abdominal ultrasound showed a hypoechoic mass, (11×9 cm) in the posterior pelvis of the bladder. Further imaging with thoracic and abdominal computed tomography (CT) scans demonstrated multiple lung nodules consistent with metastatic disease, as well as heterogeneous soft tissue masses in the abdominal cavity. Positron emission tomography/CT confirmed hypermetabolic lymphadenopathy in the mediastinum, abdomen, and pelvis, along with skeletal metastases and hypometabolic ascitic fluid. A biopsy of the abdominal mass was performed, revealing small round blue cells embedded in a desmoplastic stroma, consistent with DSRCT. Immunohistochemical analysis showed positivity for desmin and EMA, and negativity for CK7, CK20, myogenin, MyoD1, and p63. The Ki-67 proliferation index was approximately 15-20% (Table 1, Figures 1, 2).

The patient, who received a pathological diagnosis of DSRCT based on biopsy results was referred to the medical oncology department. The patient developed post-renal acute kidney injury (AKI) due to pressure from the mass during follow-up. The AKI condition was corrected by placing a nephrostomy. It was considered that the existing hypertensive condition might be related to renal artery compression. The VAC-IE chemotherapy protocol was initiated. After the first cycle of the VAC-IE regimen, the patient developed a deterioration in general condition; therefore, the treatment was continued with the VAC regimen. After the third cycle, the patient was reported to have stable disease on an interim evaluation, and the treatment was extended to the sixth cycle. At the end of the 6th cycle, the patient is still being monitored with stable disease. Follow-up appointments are ongoing.

DISCUSSION

The current case presents widespread metastasis upon initial diagnosis, which contrasts with some reported cases, where the disease is localized or regional [5]. Early diagnosis and management are of importance, given the aggressive nature of DSRCT and the poor prognosis associated with metastatic disease [6]. Given the diagnostic complexity and the need for specialized expertise in managing this rare malignancy, a multidisciplinary approach involving oncologists, surgeons, and radiologists is essential for optimizing patient outcomes.

DSRCT can be confused with other abdominal tumors such as lymphoma, rhabdomyosarcoma, neuroblastoma, primitive neuroectodermal tumor, small cell mesothelioma, Ewing's sarcoma, and Wilm's tumor, especially in young patients [7]. DSRCT is typified by a proliferation of small, round, hyperchromatic neoplastic cells encased within a densely fibrotic, desmoplastic stromal component [8]. Immunohistochemical staining is critical for confirming the diagnosis and differentiating DSRCT from

Table 1. The patient's laboratory results at presentation

Urea	32 mg/dL (7-20 mg/dL)
Creatinine	1.85 mg/dL (0.6-1.3 mg/dL)
Uric acid	8.6 mg/dL (3.5-7.2 mg/dL)
Total protein	69 g/L (64-83 g/L)
Albumin	44 g/L (3550 g/L)
Aspartate aminotransferase (AST)	22 U/L (10-40 U/L)
Alanine aminotransferase (ALT)	24 U/L (7-56 U/L)
Alkaline phosphatase (ALP)	80 U/L (40-130 U/L)
Gamma-glutamyl transferase (GGT)	32 U/L (9-48 U/L)
Lactate dehydrogenase (LDH)	304 U/L (125-250 U/L)
International normalized ratio (INR)	1.4 (0.8-1.2)
White blood cell count (WBC)	9.07x10^9/L (3.9-10.2 x10^9/L)
Neutrophil count	7.15x10^9/L (1.5-7.5 x10^9/L)
Hemoglobin	12.5 g/dL [13.5-17.5 g/dL (man), 12.0-15.5 g/dL (woman)]
Platelet count	337 x10^9/L (150-450 x10^9/L)
Carcinoembryonic antigen (CEA)	0.7 ng/mL (<3 ng/mL)
Cancer antigen 19-9 (CA 19-9)	10.7 U/mL (<37 U/mL)
Alpha-fetoprotein (AFP)	<1.3 µg/L (<10 µg/L)
Erythrocyte sedimentation rate (ESR)	35 mm/hour [<20 mm/hour (man), <30 mm/hour (woman)]
C-reactive protein (CRP)	41.10 mg/L (<5 mg/L)

other tumors [9]. DSRCT typically shows positivity for desmin, WT1, and epithelial markers such as epithelial membrane antigen and cytokeratin. In most cases, DSRCTs resemble disseminated carcinomatoses in their clinical manifestation as well as cytomorphologically, even in young adult patients [9]. Fibrillary stromal fragment, clinical setting, and adjunctive immunocytochemical staining are most helpful for avoiding misdiagnosis [9].

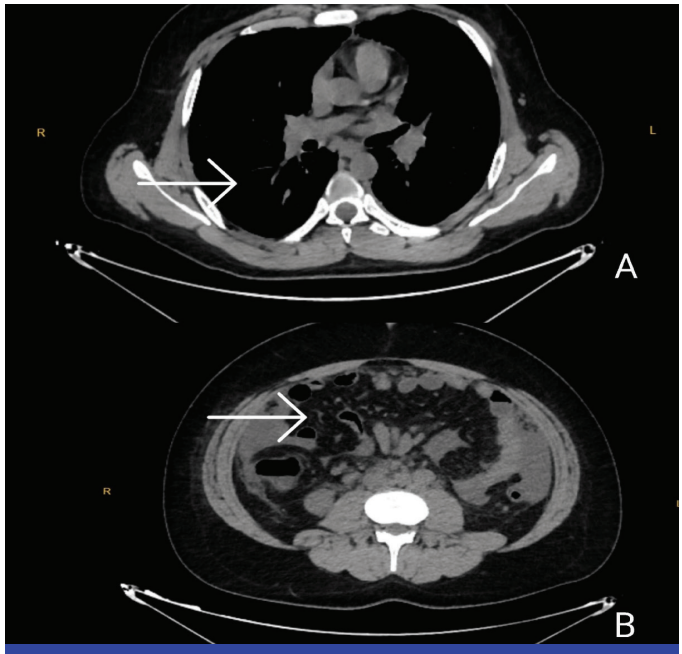

Figure 1. (A, B) The patient's thorax-abdomen CT scan arrows indicate metastases

CT: Computed tomography

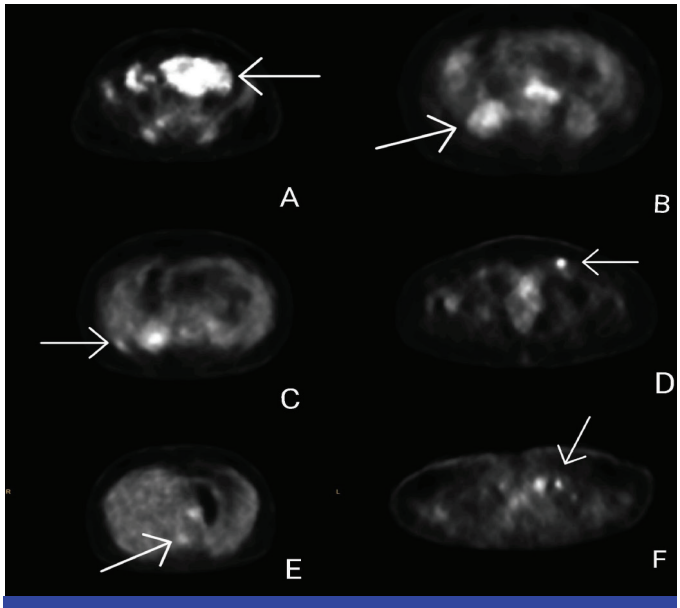

Figure 2. (A-F) The patient's PET/CT scan arrows indicate hypermetabolic lesions

PET: Positron emission tomography, CT: Computed tomography

Multimodal treatment strategies, including intensive combination chemotherapy, aggressive surgical resection, and adjuvant radiotherapy, have been the mainstay of DSRCT management and are associated with improved overall survival, according to recent studies. Recent studies have highlighted the role of Hyperthermic Intraperitoneal Chemotherapy and whole-abdomen radiotherapy in improving local disease control in patients with DSRCT [2,3]. The role of tyrosine kinase inhibitors such as pazopanib and sunitinib, which target platelet-derived growth factor receptor alpha (PDGFRα) and vascular endothelial growth factor (VEGF), has been explored in clinical trials, with some evidence of modest activity in patients with advanced DSRCT [10]. Clinical trials have assessed the efficacy of mTOR inhibitors in advanced and recurrent disease; some research trials in soft-tissue sarcoma and Ewing sarcoma include DSRCT patients, but few studies have been tailored to the specific clinical needs and underlying cytogenetic abnormalities characterizing this disease, such as the typical *EWSR1-WT1* gene fusion [2]. The presence of the EWSR1-WT1 translocation, which results in the upregulation of PDGFRα and VEGF, underscores the potential of targeted therapies in this disease [10]. Early clinical trials with IGF-1R inhibitors and PD-1/PD-L1 checkpoint inhibitors have shown promise, but further studies are needed to establish their efficacy in DSRCT [2]. The prognosis for DSRCT remains poor, with overall survival rates ranging from only 15% to 30% at five years, according to recent studies [7]. DSRCT is frequently diagnosed at an advanced stage, with widespread metastasis to the liver, lungs, and bones. The role of targeted therapies and immunotherapy in DSRCT is an area of ongoing research, and clinical trials are needed to evaluate the efficacy of these agents.

DSRCT remains a diagnostic challenge, particularly in young male patients in whom more common malignancies such as lymphoma and germ cell tumors are initially considered. This case highlights the importance of broad differential diagnoses when evaluating patients with an abdominopelvic mass, as uncommon malignancies can present similarly to more prevalent ones. The unexpected diagnosis of DSRCT underscores the necessity of a thorough diagnostic approach, including histopathological and molecular analyses, to accurately classify rare tumors. Early diagnosis and comprehensive treatment planning, incorporating multimodal therapies such as chemotherapy, surgery, and radiation therapy, remain essential for improving survival outcomes. As advancements in targeted therapies and immunotherapy continue to evolve, further research and clinical trials are warranted to explore novel therapeutic strategies that may enhance prognosis in patients with DSRCT.

Ethics

Informed Consent: Informed consent was obtained from the patient for publication of this case report, including any accompanying images and data.

Footnotes

Authorship Contributions

Surgical and Medical Practices: Y.S.T., E.S.S., A.B., B.E., En.S.Ş., Concept: Y.S.T., A.B., B.E., Design: Y.S.T., Data Collection or Processing: Y.S.T., E.S.S., A.B., Analysis or Interpretation: Y.S.T., E.S.S., Literature Search: Y.S.T., E.S.S., B.E., Writing: Y.S.T., E.S.S., A.B., B.E., En.S.Ş.

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CASE REPORT

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Effect of Hypothyroidism Treatment on a Patient with Atrial Fibrillation

Leyla Abbasova, Aygun Huseynova, Natavan Ismayilova

Azerbaijan State Advanced Training Institute for Doctors named after A. Aliyev, Department of Therapy, Baku, Azerbaijan

ABSTRACT

This case report highlights the interplay between thyroid dysfunction and atrial fibrillation (AF). While hyperthyroidism is a well-established risk factor for AF, the relationship between hypothyroidism and AF remains less understood. The case study offers insight into how thyroid hormone replacement therapy (levothyroxine) can influence AF progression and symptom control.

Keywords: Atrial fibrillation, Thyroid disfunction, Hypothyroidism

INTRODUCTION

Thyroid dysfunction is prevalent worldwide [1]. According to data, the prevalence of thyroid diseases in Europe and the United States is approximately 6.6% [2-5]. Literature reports indicate that both hyperthyroidism and hypothyroidism have adverse effects on cardiovascular health [6]. Hyperthyroidism is a strong and independent risk factor for atrial fibrillation (AF) [7]. However, the pathological mechanisms linking hypothyroidism and AF remain unclear [8]. Abnormally high thyroid hormone levels can disrupt the electrical signals regulating heart rhythm, potentially leading to AF. Even in euthyroid individuals, higher free T4 levels are associated with an increased risk of AF [8]. Based on this, we present a clinical case of a patient with AF and hypothyroidism.

CASE REPORT

A 54-year-old female was referred to the department of therapy with complaints of fatigue and heart palpitations. One year earlier, she had been diagnosed with paroxysmal tachysystolic AF: CHA_2DS_2VA-1, EHRA-3. The patient experienced four to five paroxysmal episodes per month and was taking propafenone for symptom control.

Echocardiography revealed the following parameters: left ventricular end-diastolic diameter - 50 mm, left ventricular end-systolic diameter - 35 mm, interventricular septum - 12 mm, left ventricular posterior wall - 11 mm, left atrial volume index - 32 mm/m^2, ejection fraction - 57%.

Laboratory analysis showed: thyroid stimulating hormone (TSH) - 8.41 (reference: 0.3-4 mIU/L), free T4 - 1.15 (reference: 10-25 pmol/L), free T3 - 3.76 (reference: 2.5-5.8 pmol/L), anti-thyroid peroxidase antibody - 1040 (reference: 2.5-30 IU/mL).

The thyroid ultrasound examination reported the following findings: right lobe - 18×23×39 mm; left lobe - 22×20×38 mm; isthmus thickness - 3.8 mm; total thyroid volume - 19.5 mL; echotexture - heterogeneous; vascular supply - normal; no nodules detected.

The patient began 50 mcg levothyroxine therapy. After six weeks of treatment, the TSH level improved to 3.8 mIU/L, and the patient's symptoms of fatigue and palpitations significantly decreased, with no new AF paroxysms recorded.

DISCUSSION

Although hyperthyroidism is a well-known cause of AF due to increased adrenergic activity, enhanced automaticity, and shortened atrial refractory periods, hypothyroidism is traditionally associated with bradycardia, diastolic dysfunction, and reduced cardiac output rather than AF. However, recent studies suggest that subclinical and overt hypothyroidism may contribute to arrhythmogenesis through changes in cardiac electrophysiology, autonomic regulation, and endothelial

Address for Correspondence: Aygun Huseynova MD, Azerbaijan State Advanced Training Institute for Doctors named after A. Aliyev, Department of Therapy, Baku, Azerbaijan
E-mail: ayhuseynova@gmail.com **ORCID ID:** 0000-0001-7754-9921
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function; higher TSH levels are linked to inflammation and oxidative stress, which can indirectly influence atrial remodeling and AF susceptibility.

This case further supports the idea that hypothyroidism-related AF may be underrecognized in clinical practice.

This case underscores the potential reversibility of AF in patients with hypothyroidism and highlights the importance of thyroid function assessment in AF management. Further research is needed to define the exact pathophysiological mechanisms and optimal treatment strategies for AF patients with thyroid dysfunction.

Ethics

Informed Consent: Written informed consent was obtained from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: L.A., A.H., N.I., Concept: L.A., A.H., N.I., Design: L.A., A.H., N.I., Data Collection or Processing: L.A., A.H., N.I., Analysis or Interpretation: L.A., A.H., N.I., Literature Search: L.A., A.H., N.I., Writing: L.A., A.H., N.I.

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CASE REPORT

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Good Syndrome Associated Enteropathy: A Case Report

Narmin Naghizada[1], Tuğba Tolu[2], Haluk Tarık Kani[2,3]

[1]Marmara University Faculty of Medicine, Department of Internal Medicine, İstanbul, Turkey
[2]Marmara University Faculty of Medicine, Department of Gastroenterology, İstanbul, Turkey
[3]Marmara University Faculty of Medicine, Institute of Gastroenterology, İstanbul, Turkey

ABSTRACT

Good syndrome (GS) is a rare, late-onset primary immunodeficiency disorder characterized by the presence of thymoma and hypogammaglobulinemia, with no familial inheritance. Immunological abnormalities in GS include defects in both humoral and cellular immunity. We present the case of a patient with GS who developed chronic diarrhea. Unlike other reported cases of GS-associated enteropathy in the literature, this case exhibited similarities to common variable immunodeficiency-related enteropathy. The patient's chronic diarrhea was unresponsive to intravenous immunoglobulin therapy but showed significant improvement with steroids and infliximab. This case highlights the potential for atypical presentations of GS-associated enteropathy and underscores the importance of considering alternative therapeutic strategies, including immunomodulatory agents, in refractory cases.

Keywords: Colitis, Good syndrome, Infliximab

INTRODUCTION

Good syndrome (GS) is a primary immunodeficiency disorder characterized by the association of thymoma and hypogammaglobulinemia [1]. Although it was classified as a subset of common variable immunodeficiency (CVID) in 2005 [2], GS is considered a distinct late-onset primary immunodeficiency with low peripheral B-cell counts, adult onset (typically between ages 40-60), and no familial history [3]. The International Union of Immunological Societies and the World Health Organization have acknowledged it as a separate clinical entity [4].

CASE REPORT

A 54-year-old male presented to the gastroenterology outpatient clinic with persistent diarrhea. The patient reported persistent diarrhea for 3 months; up to 10 bowel movements per day, without blood or mucus. Additionally, he lost 25 kg in the last year. In his medical history, he had a thymectomy two years ago due to thymoma. In physical examination, he was dehydrated; other than that, his physical examination was normal. The patient was hospitalized with these findings.

Initial laboratory tests: white blood cell: 8100/mL, neutrophils: 6800/mL, lymphocytes: 900/mL, hemoglobin: 11.4 g/dL, platelets: 456,000/mL, creatinine: 0.43 mg/dL, aspartate aminotransferase: 13 U/L, alanine aminotransferase: 12 U/L, albumin: 28 g/L, total protein: 43 g/L, procalcitonin: 0.17 mg/L, C-reactive protein: 95 mg/L. There was no pathological finding in ileo-colonoscopy, and multiple biopsies were performed from each segment of the terminal ileum and colon.

Laboratory results showed significantly low immunoglobulin (Ig) levels: IgG 1.19 g/L, IgM <0.2 g/L, and IgA <0.1 g/L. Isohemagglutinin testing revealed anti A-IgM negativity and anti B-IgM positivity. Hypogammaglobulinemia, the presence of thymoma, and recurrent infections led to a diagnosis of GS. Intravenous immunoglobulin (IVIG) therapy was scheduled to be administered every 21 days.

Hypoxemia (oxygen saturation: 85%) and tachypnea (respiratory rate: 32/min) developed on the 7th day of hospitalization. A chest computed tomography scan revealed bilateral ground-glass opacities. Sputum cultures and a respiratory viral panel were sent for analysis. Opportunistic infections, including tuberculosis, Pneumocystis jirovecii pneumonia (PCP), and cytomegalovirus (CMV)

Address for Correspondence: Haluk Tarık Kani MD, Marmara University Faculty of Medicine, Department of Gastroenterology; Marmara University Faculty of Medicine, Institute of Gastroenterology, İstanbul, Turkey
E-mail: tarik.kani@marmara.edu.tr **ORCID ID:** 0000-0003-0042-9256
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pneumonia, were considered in the differential diagnosis. He was started on intravenous trimethoprim/sulfamethoxazole (15 mg/kg) and methylprednisolone for suspected PCP. However, the bronchoalveolar lavage (BAL) PCP test result was negative, and the empirical treatment was discontinued. HIV and tuberculosis tests were negative. Plasma CMV polymerase chain reaction (PCR) was 1,096 copies/mL, while BAL CMV PCR showed 299,168 copies/mL; confirming CMV pneumonia. Additionally, the colonoscopy revealed CMV colitis. Intravenous ganciclovir (5 mg/kg twice daily) was initiated, resulting in symptom improvement. After this, the patient was transitioned to oral valganciclovir for maintenance therapy. The patient was discharged and continued his follow-up in the immunology outpatient clinic.

Four months later, the patient presented with diarrhea again (15-20 bowel movements per day). Stool culture, Giardia, Entamoeba histolytica, Clostridium difficile antigen, and parasites in stool were negative; and repeat colonoscopy ruled out CMV colitis. New biopsies showed findings (such as a reduction in plasma cells and eosinophilia) consistent with GS-associated enteropathy. Oral methylprednisolone (32 mg) was started, resulting in temporary symptom relief. However, diarrhea recurred (8-10 episodes daily) as the steroid dose was tapered. Stool testing revealed Clostridium difficile antigen positivity. The patient was treated with oral vancomycin (125 mg four times daily) and metronidazole (500 mg three times daily), which together resolved his symptoms. Despite this, the diarrhea relapsed upon further steroid tapering.

Despite initial improvements, the patient experienced a relapse of colitis upon further steroid tapering. Following a consultation with the Allergy and Immunology Department, intravenous infliximab therapy (5 mg/kg every 8 weeks) was initiated. This resulted in complete remission, and the patient was referred for follow-up in both gastroenterology and immunology clinics.

DISCUSSION

Enteropathy is a common manifestation in primary immunodeficiency disorders. It can present with chronic diarrhea, malabsorption, growth delay, iron deficiency anemia, and other symptoms of malnutrition. The most common causes are celiac disease (26.2%), Immunodysregulation Polyendocrinopathy Enteropathy X-linked syndrome (20.7%), autoimmune enteropathy (6.4%), and CVID-associated enteropathy (5.8%) [5].

A recent study reviewed on GS cases analyzing 225 patients was published between November 2020 and October 2022, among them, 22 (9.8%) had non-infectious gastrointestinal involvement, including inflammatory bowel disease (n=4), multiorgan autoimmunity involving the gastrointestinal system (n=3), celiac disease (n=1), collagenous colitis (n=1), autoimmune enteropathy (n=1), and non-granulomatous colitis (n=1). Chronic diarrhea (90.9%) and significant weight loss (50.0%) were the most common clinical findings [6].

Clinical improvement was observed in 11 patients with IVIG, and two patients improved following thymectomy [6].

In contrast, our case demonstrated no response to IVIG but achieved remission with infliximab, similar to the treatment approach for CVID-associated enteropathy [7].

GS remains a rare clinical entity, with no established consensus regarding its treatment or pathophysiology. Chronic diarrhea is a common symptom of this syndrome. Once infectious causes are excluded, further research and case series are essential to better understand immune-mediated enteropathies associated with GS.

Ethics

Informed Consent: A written informed consent has been granted from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: H.T.K., Concept: N.N., T.T., H.T.K., Design: N.N., T.T., H.T.K., Data Collection or Processing: N.N., T.T., H.T.K., Analysis or Interpretation: N.N., T.T., H.T.K., Literature Search: N.N., T.T., H.T.K., Writing: N.N., T.T., H.T.K.

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CASE REPORT

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A Rare Cause of Abdominal Pain and Diarrhea-cystic Lymphangioma Located in the Descending Colon

Wei Yang[1], Chendong He[2]

[1]Jiangsu Province Hospital of Chinese Medicine and Affiliated Hospital of Nanjing University of Chinese Medicine, Department of Radiology, Nanjing, China
[2]Nanjing Hospital of Chinese Medicine Affiliated to Nanjing University of Chinese Medicine, Department of Radiology, Nanjing, China

ABSTRACT

Cystic lymphangiomas of the colon are rare, benign tumors that arise from the lymphatic vessels and are typically asymptomatic. These lesions are often discovered incidentally during imaging or endoscopic evaluations for unrelated conditions. We report a case of a 57-year-old woman who presented with abdominal pain and diarrhea. Imaging studies revealed a submucosal cystic mass in the descending colon, and endoscopic evaluation confirmed the presence of a cystic lesion. Histopathological examination following biopsy confirmed the diagnosis of cystic lymphangioma. The lesion was successfully resected via colonoscopy, and the patient had an uneventful recovery. This case highlights the importance of considering cystic lymphangiomas in the differential diagnosis of gastrointestinal masses and underscores the value of endoscopic techniques for diagnosis and treatment.

Keywords: Endoscopic submucosal dissection, Colon, Cystic lymphangioma

INTRODUCTION

Lymphangiomas are rare, benign tumors originating from the lymphatic vessels, most commonly found in the skin and soft tissues. Although they can occur anywhere in the body, their presence in the gastrointestinal tract, particularly in the colon, is extremely rare [1]. Cystic lymphangiomas are often asymptomatic and are frequently discovered incidentally during diagnostic imaging or endoscopy for other gastrointestinal issues. When symptomatic, they can present with a variety of non-specific symptoms such as abdominal pain, bloating, or gastrointestinal bleeding. In severe cases, complications such as perforation or intestinal obstruction can occur.

Diagnosis is generally made through imaging studies such as ultrasound, computed tomography (CT), or magnetic resonance imaging, which reveal characteristic cystic masses. However, a definitive diagnosis requires a histopathological examination of tissue obtained via biopsy, which demonstrates the characteristic lymphatic spaces lined by endothelial cells. Treatment of cystic lymphangiomas in the colon is typically surgical or endoscopic. While small, asymptomatic lesions may be observed, larger, or symptomatic lesions usually require resection. Colonoscopy provides a minimally invasive method for both diagnostic evaluation and therapeutic intervention.

CASE REPORT

A 57-year-old female patient was admitted with a chief complaint of lower abdominal discomfort, present for over one year and worsened over the past week, without any identifiable precipitating factors. She denied associated symptoms such as nausea, vomiting, or melena and did not seek medical attention at that time. One week before admission, the abdominal discomfort acutely worsened and was accompanied by diarrhea. The patient had no known history of hypertension, coronary artery disease, or diabetes, and denied any prior history of infectious diseases, including hepatitis and tuberculosis. On physical examination, the abdomen was soft and non-tender, with no evidence of abdominal wall varicosities, rebound tenderness, fluid thrill, succussion splash, or palpable masses. Bowel sounds were within normal limits. Laboratory investigations, including complete blood count,

Address for Correspondence: Chendong He MD, Nanjing Hospital of Chinese Medicine Affiliated to Nanjing University of Chinese Medicine, Department of Radiology, Nanjing, China
E-mail: hcd1222@163.com **ORCID ID:** https://orcid.org/0000-0002-2216-7245
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renal function tests, coagulation profile, cardiac enzyme panel, troponin, alpha-fetoprotein, carcinoembryonic antigen, and carbohydrate antigen 19-9, were unremarkable.

Contrast-enhanced CT of the abdomen revealed a cystic lesion located within the submucosal layer of the descending colon (Figure 1a and b). The patient subsequently underwent endoscopic submucosal dissection (ESD). Intraoperatively, a translucent submucosal elevation was observed in the descending colon, characterized by a soft consistency and smooth surface (Figure 1c). The lesion was completely resected, measuring approximately 3 cm×2 cm. Histopathological analysis demonstrated a cystic structure within the submucosa, partially septated and lined by a single layer of flattened endothelial cells, consistent with a diagnosis of cystic lymphangioma (Figure 2).

On the second postoperative day, the patient experienced a temperature peak of 39 °C, along with elevated blood levels of hypersensitive C-reactive protein (26.65 mg/L). Additionally, the white blood cell count was 11.44×10^9/L with a neutrophil percentage of 90.4%. Despite treatment with the anti-infective agent ornidazole, no significant improvement was observed. Another abdominal CT scan was performed, which revealed the presence of gas and exudation within the abdomen. Consequently, a bowel perforation was diagnosed, leading to the decision to perform an emergency laparotomy and temporary colostomy. At the two-week postoperative follow-up, the surgical wound had healed well and the patient was asymptomatic, discharged in stable condition.

DISCUSSION

Lymphangiomas are rare benign lesions, with approximately 95% of cases occurring in the head and neck region. Abdominal lymphangiomas are commonly found in the mesentery and retroperitoneum. Lymphangiomas involving the bowel wall are less frequent, accounting for around 0.7% of all abdominal lymphangiomas, and they typically manifest in the right colon. In adults, bowel wall lymphangiomas are usually secondary and may arise as a result of lymphatic endothelial cell proliferation stimulated by inflammation or previous surgical interventions. Lymphangiomas of the colon can be broadly classified into three types: simple (capillary), spongy, and cystic [2], with the cystic subtype representing approximately 70% of cases. Clinical manifestations of cystic lymphangioma of the colon are non-specific and commonly include abdominal pain, vomiting, and constipation. Complications such as intestinal obstruction, intussusception, and gastrointestinal bleeding are infrequent occurrences [3]. Endoscopic ultrasound is the preferred method for evaluating colonic cystic lymphangiomas. These lesions typically appear as submucosal, well-circumscribed, completely encapsulated cysts without echogenicity. CT imaging commonly reveals a watery cystic mass with thin walls, showing no significant enhancement on contrast-enhanced

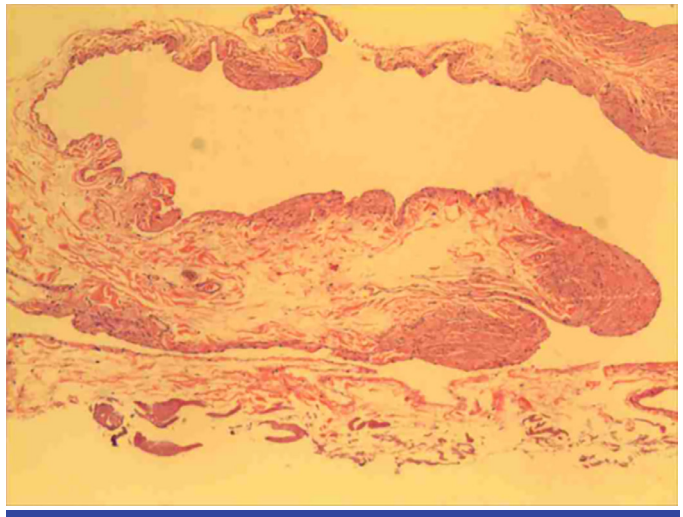

Figure 2. Pathological findings. A cystic structure was observed within the submucosa, with partial septation in some areas. The cysts were lined by a single layer of flattened endothelial cells, consistent with a diagnosis of cystic lymphangioma

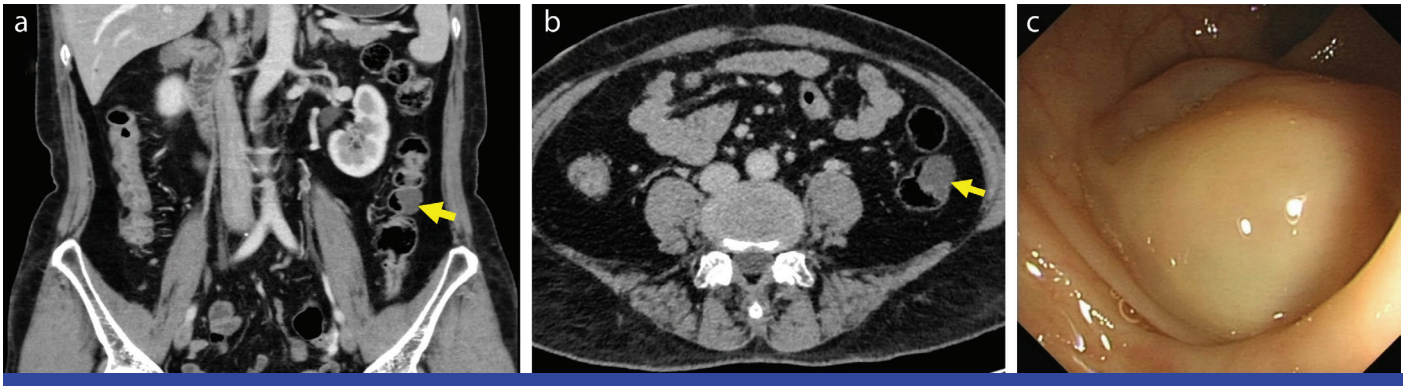

Figure 1. CT and endoscopy findings. a) Coronal and (b) axial CT images revealed a cystic lesion (yellow arrow) in the wall of the descending colon; (c) a submucosal eminence in the colon with a smooth and transparent surface

CT: Computed tomography

scans. However, they can resemble other cystic foci in the colon, which must be distinguished to avoid misdiagnosis. Mesenteric cysts, though well-circumscribed and fluid-filled like lymphangiomas, are located outside the colon wall and are often attached to the mesentery, differentiating them from intramural cysts. Gastrointestinal stromal tumors may also show cystic changes but are usually located in the muscularis propria, whereas lymphangiomas arise from the submucosa. Additionally, congenital intestinal duplication cysts are typically lined by gastrointestinal epithelium, a feature absent in lymphangiomas. Infectious or inflammatory cystic lesions, such as abscesses or granulomas, may present with more heterogeneous features and associated systemic symptoms. Accurate diagnosis often requires a combination of clinical assessment, imaging, and histopathological examination to confirm the benign nature of cystic lymphangioma and rule out other conditions. Colonic cystic lymphangioma is a rare benign malformation, and the decision for active intervention depends on clinical symptoms. Endoscopic resection is a suitable approach for small lesions measuring 2 to 3.5 cm [4]. However, it is essential to be cautious about the potential complications of colon perforation.

In the present case, colon perforation occurred after ESD, necessitating urgent surgical intervention. Laparoscopic surgery may be considered as an alternative to mitigate complications such as recurrent infection, progressive growth, rupture, or bleeding.

Ethics

Informed Consent: Informed consent was obtained from the patient for the anonymous use and publication of clinical and imaging data.

Footnotes

Authorship Contributions

Concept: W.Y., C.H., Design: W.Y., C.H., Data Collection or Processing: W.Y., C.H., Analysis or Interpretation: W.Y., C.H., Literature Search: W.Y., C.H., Writing: W.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

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