CASE REPORT
Turk J Surg

Early-onset colorectal cancer in pediatric patient

Sarp Tunalı[1], Kamil Erözkan[1] , Yiğit Türk[1], Ülgen Çeltik[2], Osman Bozbıyık[1]

[1]Department of General Surgery, Ege University Hospital, İzmir, Türkiye
[2]Department of Pediatric Surgery, Ege University Hospital, İzmir, Türkiye

ABSTRACT

The incidence of early-onset colorectal cancer is rising, largely driven by underlying genetic mutations. While many countries have implemented screening programs for individuals under 45 years of age, colorectal cancer in pediatric patients remains exceedingly rare and often unexpected. As a result, children diagnosed with colorectal cancer frequently present with advanced-stage disease, commonly due to obstructive symptoms. In this case report, we describe the clinical presentation and management of a 17-year-old female patient who was admitted with an obstructing tumor at the splenic flexure.A 17-year-old girl with cerebral palsy was referred to our center with a 10-day history of obstructive symptoms. She had a four-month history of constipation with multiple hospital admissions. Following an evaluation by the pediatric surgery team in the emergency department, a computed tomography scan was performed, revealing a 4 cm tumor at the splenic flexure with signs of obstruction and suspected metastatic mesenteric lymphadenopathy. Once the diagnosis was confirmed, the pediatric surgery team referred the patient to the colorectal surgery team. The colorectal surgeons recommended laparotomy, which was performed through a midline incision. An extended left hemicolectomy with a Mikulicz colostomy was carried out. The patient was discharged on postoperative day 7 without complications. Histopathological analysis revealed mucinous adenocarcinoma with 17 metastatic lymph nodes out of 42 harvested. No BRAF, KRAS, or NRAS mutations were detected; mismatch repair protein expression was intact. Genetic counseling identified a variant of uncertain significance in the *ATM* gene. The patient was subsequently referred for adjuvant chemotherapy. Pediatric-onset colorectal cancer should be considered in the differential diagnosis of children presenting with persistent gastrointestinal symptoms, as delayed recognition may lead to advanced disease. Genetic counseling plays a crucial role in management, underscoring the need for surgeon awareness and multidisciplinary collaboration.

Keywords: Cancer, colorectal cancer, gastrointestinal surgery, general surgery

INTRODUCTION

Colorectal carcinoma is the 3rd most common type of cancer worldwide. Its mortality is decreasing due to recently developed and more accessible diagnostic methods. Regarding the age range, colon carcinoma is rare in pediatric patients. Current data indicate that it constitutes 1% of pediatric neoplasms (1). This rarity is actually due to the lack of awareness of genetic diseases. Although symptoms such as anemia and changes in bowel habits seen in early-onset colorectal cancers (CRCs) show similarities across all age groups, in young patients, these findings are frequently attributed to benign conditions. This situation, combined with a lack of clinical suspicion, leads to significant delays in the diagnostic process and the diagnosis of the disease at advanced stages. The prognosis remains poor even in patients who undergo radical surgery. This case report aims to present a 17-year-old patient who had signs of obstruction due to colon cancer and was operated on for this reason.

CASE REPORT

A 17-year-old female patient with a known diagnosis of cerebral palsy applied to the pediatric surgery department of our center with a history of persistent constipation-diarrhea (change in bowel habits) lasting for the last 4 months, obstructive symptoms continuing for 10 days, anorexia, inability to defecate for 8 days, and nausea and vomiting for the last 3 days. She had multiple hospital admissions in the last six months. A diagnosis could not be made for the patient during these admissions. Routine biochemistry and hemogram tests and plain abdominal X-ray (ADBG) tests had been performed on the patient at an external center. In these tests performed, although no additional pathology was seen on her ADBG, minimal anemia was present in her blood tests. When the patient, who was consulted to us by pediatric surgery, was evaluated; her vital signs were observed to be stable and she had no fever. There had been no stool output for approximately 8 days.

Cite this article as: Tunalı S, Erözkan K, Türk Y, Çeltik Ü, Bozbıyık O. Early-onset colorectal cancer in pediatric patient *Turk J Surg*. [Epub Ahead of Print]

Corresponding Author
Kamil Erözkan

E-mail: kamilerozkan@gmail.com
ORCID ID: orcid.org/0000-0003-2193-9984

Received: 02.11.2025
Accepted: 17.02.2026
Epub: 24.02.2026

DOI: 10.47717/turkjsurg.2026.2025-11-4

Available at **www.turkjsurg.com**

On examination, although there were no signs of guarding, rebound, or acute abdomen, distension was present. Digital rectal examination was empty. Metallic sounds were heard on auscultation. In her blood tests seen in our clinic, C-reactive protein was 72 mg/L, white blood cell 7.14 10^3/µL, procalcitonin 0.66 µg/L, hemoglobin 11.4 g/dL, hematocrit 35.3%, platelet 400 10^3/µL, liver function tests, creatinine and electrolyte values were within normal range. After all these findings, it was decided to perform a computed tomography scan on the patient. In the computed tomography examination performed, a 4 cm mass causing obstruction at the splenic flexure and mesenteric lymphadenopathies thought to be metastatic were detected (Figures 1 and 2). The case was referred to colorectal surgery by the pediatric surgery team who made the initial evaluation. A decision for surgery was made for the patient with obstruction findings. In the laparotomy performed with a midline incision, the obstructive tumor was seen (Figure 3) and an extended left hemicolectomy + Mikulicz colostomy operation was performed. Due to the patient being in a picture of acute mechanical intestinal obstruction, her nutritional status progressing suboptimally, and postoperative care difficulties due to cerebral palsy, extended left hemicolectomy and Mikulicz colostomy were preferred in order to minimize the risk of anastomotic leakage. No intraoperative signs of peritoneal carcinomatosis or intra-abdominal metastasis findings were seen. Metastatic lymph nodes were seen in the mesentery and these were excised according to oncological cancer surgery principles. The patient was discharged without complications on the postoperative 7th day. Mucinous adenocarcinoma was observed in the histopathological examination; metastases were detected in 17 of the 42 lymph nodes examined. Surgical margins were reported as negative. According to TNM staging, it was pT4 N2 M0. The case was staged as Stage IIIC according to American Joint Committee on Cancer 8th edition. R0 resection was achieved. In advanced pathological examinations, no mutations were detected in *BRAF* gene Codon 600, *NRAS* Gene EXON 2 (Codon 12 and 13), EXON 3 (Codon 61), *KRAS* gene, EXON 2 (Codon 12 and 13), EXON 3 (Codon 61). *cerbB2* was detected as focal 2 (+) positive. No loss in DNA mismatch repair protein expression was detected. As a result of genetic counseling, a variant of unknown significance (VUS) was detected in the *ATM* gene. The patient

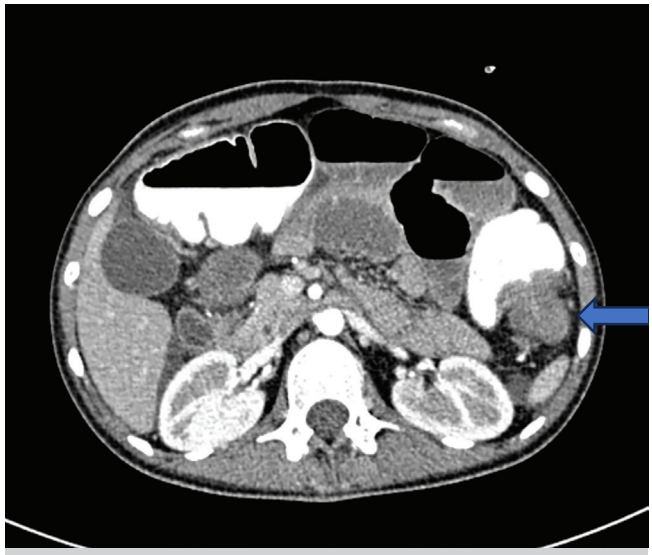

Figure 2. Preoperative axial CT image.
CT: Computed tomography.

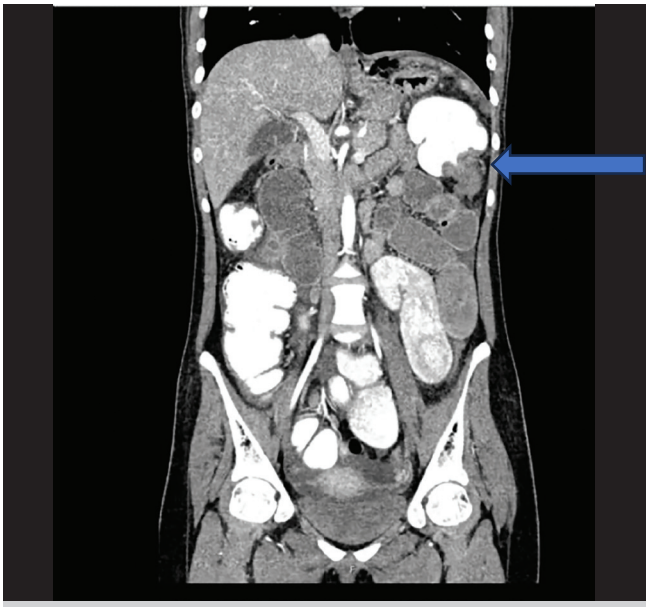

Figure 1. Preoperative coronal CT image.
CT: Computed tomography.

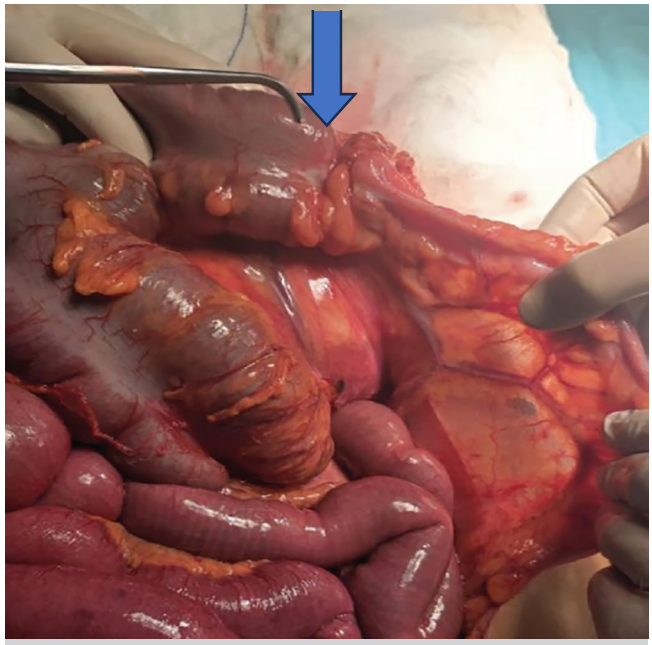

Figure 3. Intraoperative view of the surgical field.

and her relatives were referred to the genetic diseases unit. The patient was referred for adjuvant chemotherapy with the test results.

DISCUSSION

CRC is rare in children and adolescents. It constitutes 1% to 2% of all pediatric tumors. It's incidence is estimated to be 1/1,000,000 (1). In pediatric cases, the frequency of occurrence in males is approximately twice as much compared to females (2). In recent years, the incidence of early-onset CRC has been increasing in many countries (3-5). In the United States of America, the incidence has risen since the 1990s; while it was 5.9 per 100,000 in 2000, it rose to 8.4 in 2017 (6). Similar increases have also been reported in Europe, and early-onset CRC has become a significant cancer burden among young adults today (3).

Diagnosing CRC in patients with cerebral palsy involves technical and clinical challenges due to the existing neurological condition masking the symptoms. Chronic constipation and gastrointestinal motility disorders, which are common in these patients, can lead to early signs of obstruction caused by the tumor being perceived as a routine condition. Additionally, communication barriers increase diagnostic delay by making it difficult for patients to report alarm symptoms. Studies have reported that motor function impairment in cerebral palsy contributes to an increased risk of cancer; while physical exercise is protective against the development of colon cancer (7), an increase in colon cancer has been reported in patients with cerebral palsy (8).

When looking at risk factors for early-onset CRCs, recent studies include changes in the human microbiota, Western-style diet, alcohol, tobacco use, red meat consumption, inflammatory bowel disease, family history of CRC, and genetic variants predisposing to CRC, while protective factors include aspirin use, high systemic vitamin D levels, high folate intake, and physical activity (4). No clear consensus has been established in studies regarding obesity and inactivity among these risk factors (5). When our patient was examined retrospectively, there was no family history of CRC.

When a review of previous studies is conducted, CRC is seen twice as much in boys compared to girls (2). Rectal bleeding is the most commonly reported red flag finding. Abdominal pain, changes in bowel habits, weight loss, loss of appetite, and anemia are also frequently encountered symptoms (9). In a recent study, it was found that rectal pain, abdominal pain, rectal bleeding, changes in bowel habits, and weight loss were associated with an increased risk of early-onset CRC (6). In another recent study, it was reported that iron deficiency anemia and rectal bleeding increased the risk of early-onset CRC tenfold (10).

Although red flag symptoms pose a risk in early-onset CRC, diagnosis is mostly delayed. Studies show that the average time from the onset of symptoms to diagnosis is 6 months (11,12). There are several explanations for this, but these explanations need to be corrected by the healthcare system before it is too late. One reason for diagnostic delay is that physicians perceive the possibility of malignancy as low due to the young age (13). Another issue is the misattribution of symptoms to benign conditions, the best example of which is considering a patient with rectal bleeding as having hemorrhoids; moreover, no other suspicion is raised if the patient is young. Despite the delayed diagnosis, current studies have not shown that this negatively affects the course of early-stage CRC or 5-year survival (5).

The gold standard in diagnosis is colonoscopy. Computed tomography and serum markers are also helpful. Especially the carcinoembryonic antigen level carries prognostic importance; however, it has limited value in diagnosis alone due to low sensitivity and false positivity (14).

The most effective treatment in CRC is surgical resection. Neoadjuvant therapy can be applied in metastatic cases or cases not suitable for resection; in the case of obstruction, diversion or the opening of an end stoma may be required. If the tumor shrinks after neoadjuvant therapy, the surgical option may come back to the agenda. In our patient, laparotomy was performed due to emergency obstruction. The operation was concluded with an extended left hemicolectomy and Mikulicz colostomy due to the diameter difference between the proximal and distal loops. No metastasis was detected on the peritoneal surfaces, and the lymph nodes in the mesentery were removed in accordance with cancer surgery principles. Colostomy closure is planned following the completion of adjuvant chemotherapy and the nutritional stabilization of the patient. This process can take up to one year. Detection of genetic variants such as ATM in early onset-CRC cases requires a multidisciplinary approach in terms of both patient follow-up and family screening.

While our case reflects the typical characteristics of CRC in the pediatric age group, it is also noteworthy in some aspects. In most pediatric cases reported in the literature, the histopathological subtype is mucinous adenocarcinoma, and this condition has been associated with poor prognosis (2). Similarly, mucinous adenocarcinoma was observed in our patient as well. In some cases, distinct genetic predispositions such as *CMMRD*, *MUTYH*, or *MMR* gene mutations have been defined (6). While mutations associated with Lynch syndrome and familial adenomatous polyposis have been reported more frequently in pediatric cases in the literature, the role of ATM variants is still unclear. In our case, the detection of a VUS in the *ATM* gene emphasizes the importance of genetic counseling in pediatric CRCs. Similarly, a case reported at the age of 15 also presented with constipation and obstruction findings and was diagnosed at an advanced

stage (2). In our patient, the symptoms lasting for approximately six months shows that diagnostic delay is frequently seen in pediatric cases.

CONCLUSION

CRC is one of the most common malignancies globally and has shown a significant increase in incidence, especially in the young population, in recent years. This situation reveals that the disease is not only unique to the elderly aged patient group and that early-onset cases should not be overlooked. Although extremely rare in the pediatric age group, CRC must be considered in the differential diagnosis of children presenting with persistent gastrointestinal symptoms.

Delayed diagnosis complicates surgical and oncological treatment processes and increases mortality. The approximately six-month delay in our patient's diagnosis clearly demonstrated this. The attribution of symptoms such as abdominal pain, change in bowel habits, rectal bleeding, weight loss, anorexia, and anemia to benign causes due to young age can also lead to serious diagnostic delays. Therefore, increasing the awareness of all physicians, especially family practitioners, pediatricians, and surgeons, is critically important.

Early diagnosis and rapid intervention directly affect the prognosis. A multidisciplinary approach, supporting surgical resection with appropriate adjuvant treatment and genetic counseling, are fundamental factors that improve treatment success and survival. In conclusion, it should not be forgotten that CRC can occur in the pediatric period, and every child with persistent gastrointestinal symptoms should be carefully evaluated for malignancy.

Ethics

Informed Consent: Written informed consent was obtained from the patient's legal guardian for the use and publication of clinical data for scientific purposes.

Footnotes

Author Contributions

Concept - S.T., K.E., Y.T., Ü.Ç., O.B.; Design - S.T., K.E., Y.T., Ü.Ç., O.B.; Data Collection or Processing - S.T., K.E., Y.T., Ü.Ç., O.B.; Analysis or Interpretation - S.T., K.E., Y.T., Ü.Ç., O.B.; Literature Search - S.T., K.E., Y.T., Ü.Ç., O.B.; Writing - S.T., K.E., Y.T., Ü.Ç., O.B.

Conflict of Interest: No conflict of interest was declared by the authors. One of the authors of this article (Y.T.) is a member of the Editorial Board of this journal. He was completely blinded to the peer review process of the article.

Financial Disclosure: The authors declared that this study received no financial support.

REFERENCES

1. *Bray F, Laversanne M, Sung H, Ferlay J, Siegel RL, Soerjomataram I, et al. Global cancer statistics 2022: GLOBOCAN estimates of incidence and mortality worldwide for 36 cancers in 185 countries. CA Cancer J Clin. 2024;74:229-263.*
2. *Abbaoui S, Zaari N, Ammor A, Benhaddou H. Adenocarcinoma of the colon in children with LAL: a case report. Int J Surg Case Rep. 2024;121:109995.*
3. *Vuik FE, Nieuwenburg SA, Bardou M, Lansdorp-Vogelaar I, Dinis-Ribeiro M, Bento MJ, et al. Increasing incidence of colorectal cancer in young adults in Europe over the last 25 years. Gut. 2019;68:1820-1826.*
4. *Keum N, Giovannucci E. Global burden of colorectal cancer: emerging trends, risk factors and prevention strategies. Nat Rev Gastroenterol Hepatol. 2019;16:713-732.*
5. *Burnett-Hartman AN, Lee JK, Demb J, Gupta S. An update on the epidemiology, molecular characterization, diagnosis, and screening strategies for early-onset colorectal cancer. Gastroenterology. 2021;160:1041-1049.*
6. *Syed AR, Thakkar P, Horne ZD, Abdul-Baki H, Kochhar G, Farah K, et al. Old vs new: risk factors predicting early onset colorectal cancer. World J Gastrointest Oncol. 2019;11:1011-1020.*
7. *Batty D, Thune I. Does physical activity prevent cancer? Evidence suggests protection against colon cancer and probably breast cancer. BMJ. 2000;321:1424-1425.*
8. *Day SM, Brooks J, Strauss D, Shumway S, Shavelle RM, Kush S, et al. Cancer mortality in cerebral palsy in California, 1988-2002. Int J Disabil Hum Dev. 2008;7:427-434.*
9. *Eng C, Jácome AA, Agarwal R, Hayat MH, Byndloss MX, Holowatyj AN, et al. A comprehensive framework for early-onset colorectal cancer research. Lancet Oncol. 2022;23:e116-e128.*
10. *Dharwadkar P, Zaki TA, Murphy CC. Colorectal cancer in younger adults. Hematol Oncol Clin North Am. 2022;36:449-470.*
11. *Olivo R, Ratnayake S. Colorectal cancer in young patients: a retrospective cohort study in a single institution. ANZ J Surg. 2019;89:905-907.*
12. *Mauri G, Sartore-Bianchi A, Russo AG, Marsoni S, Bardelli A, Siena S. Early-onset colorectal cancer in young individuals. Mol Oncol. 2019;13:109-131.*
13. *Scott RB, Rangel LE, Osler TM, Hyman NH. Rectal cancer in patients under the age of 50 years: the delayed diagnosis. Am J Surg. 2016;211:1014-1018.*
14. *Chung MJ, Chung SM, Kim JY, Ryu MR. Prognostic significance of serum carcinoembryonic antigen normalization on survival in rectal cancer treated with preoperative chemoradiation. Cancer Res Treat. 2013;45:186-192.*