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# An Integrated Approach to MSI-High Colorectal Cancer Therapy in a Young Patient: A Clinical Case

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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**Patient:** Male, 16-year-old
**Final Diagnosis:** Colorectal cancer
**Symptoms:** Persistent abdominal pain, progressive abdominal distension
**Clinical Procedure:** —
**Specialty:** Oncology

**Objective:** Unknown etiology

**Background:** Colorectal cancer (CRC) is extremely rare in adolescents and often presents with advanced disease because of nonspecific symptoms. Deficient mismatch repair (dMMR) and microsatellite instability-high (MSI-H) are important biomarkers that influence prognosis, treatment selection, and the need for genetic evaluation. We report the case of an adolescent with MSI-H-associated CRC presenting with acute intestinal obstruction.

**Case Report:** A 16-year-old male initially underwent appendectomy for presumed acute appendicitis. Persistent postoperative bowel obstruction prompted computed tomography (CT), revealing a descending colon tumor requiring emergency Hartmann's procedure. Histopathology demonstrated mucinous adenocarcinoma, pT4N0M0, with vascular invasion. Immunohistochemistry showed loss of MSH2/MSH6 expression with retained MLH1/PMS2, indicating dMMR and findings highly suggestive of Lynch syndrome, although germline testing was unavailable. Molecular analysis identified a KRAS mutation without NRAS alterations. Given his multiple high-risk features, including T4 disease, bowel obstruction, vascular invasion, mucinous histology, and very young age, he received 12 cycles of adjuvant FOLFOX chemotherapy. Treatment was well tolerated, and he remains disease-free more than 5 years after completion of therapy.

**Conclusions:** This case highlights the diagnostic complexity of adolescent CRC and the need to consider colorectal malignancy in young patients with persistent or atypical abdominal symptoms. Routine MMR/MSI testing and comprehensive molecular profiling are essential in young-onset CRC to identify patients who may have hereditary cancer syndromes and require genetic counseling. Although limited to a single case, this report underscores the importance of multidisciplinary decision-making and individualized treatment based on clinicopathological and molecular characteristics, which may contribute to favorable long-term outcomes.

**Keywords:** Young Adult • Colonic Diseases

**Full-text PDF:** https://www.amjcaserep.com/abstract/index/idArt/953248





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## Introduction

Colorectal cancer (CRC) is the third most commonly diagnosed malignancy and the second leading cause of cancer-related mortality worldwide, accounting for more than 1.9 million new cases and approximately 935 000 deaths annually [1]. Although CRC predominantly affects older adults, its incidence among adolescents and young adults has increased steadily over the past 2 decades in many countries, making early-onset CRC an emerging public health concern [2-4]. In contrast, CRC in children and adolescents remains exceptionally rare, accounting for less than 1% of all pediatric malignancies, with an estimated incidence of only 1 to 2 cases per million children each year [5,6].

Pediatric and adolescent CRC differs substantially from adult-onset disease. Tumors diagnosed at a young age are more frequently associated with mucinous or signet-ring cell histology, poor differentiation, advanced local invasion, and metastatic presentation, all of which contribute to poorer survival outcomes [5-7]. Because the presenting symptoms—including abdominal pain, constipation, bowel obstruction, anemia, and weight loss—are nonspecific, diagnosis is frequently delayed, and the disease is often mistaken for more common gastrointestinal or surgical conditions such as appendicitis or inflammatory bowel disease [4-7]. Consequently, children and adolescents are more likely to present with locally advanced disease requiring urgent surgical intervention [6,7].

Recent advances in molecular oncology have demonstrated that early-onset CRC has distinct biological characteristics compared with sporadic CRC in older adults [3,8]. Molecular profiling has become an essential component of modern CRC management because it provides prognostic information, guides therapeutic decisions, and identifies patients with possible hereditary cancer syndromes [8-10]. Among the most clinically relevant biomarkers are deficiency of the DNA mismatch repair (MMR) system and microsatellite instability-high (MSI-H), which are present in approximately 15% to 20% of colorectal cancers [9-11]. Tumors with deficient mismatch repair (dMMR) have characteristic pathological features, including mucinous differentiation, prominent lymphocytic infiltration, and an increased mutational burden, while also demonstrating distinct responses to systemic therapy [10,11].

Loss of MMR protein expression involving MSH2 and MSH6 on immunohistochemistry is highly suggestive of an underlying hereditary mismatch repair defect and should prompt referral for genetic counseling and germline testing [12-15]. However, immunohistochemical findings alone are insufficient to establish a diagnosis of Lynch syndrome, which requires confirmation by germline molecular testing. Current international guidelines therefore recommend universal MMR/MSI testing in all newly diagnosed colorectal cancers, particularly in young patients, to facilitate identification of individuals who may have an inherited cancer predisposition syndrome and who may benefit from tailored surveillance strategies for themselves and their relatives [13-16].

The therapeutic implications of molecular profiling have expanded considerably during the past decade. Patients with stage II dMMR colon cancer generally have a favorable prognosis and derive limited benefit from fluoropyrimidine monotherapy; however, oxaliplatin-containing adjuvant chemotherapy may be considered when high-risk pathological features such as T4 disease, bowel obstruction, vascular invasion, perforation, or inadequate lymph node sampling are present [16,17]. Furthermore, MSI-H/dMMR has become one of the most important predictive biomarkers for immune checkpoint inhibitors, which have demonstrated durable clinical benefit in advanced colorectal cancer [18,19]. Assessment of additional molecular alterations, including KRAS, NRAS, and BRAF mutations, also remains an essential component of comprehensive molecular characterization because these biomarkers influence prognosis and selection of targeted systemic therapies in advanced disease [20-22].

Despite increasing recognition of early-onset CRC, reports describing colorectal cancer in adolescents remain limited. Individual case reports continue to provide valuable clinical evidence by illustrating diagnostic challenges, pathological characteristics, molecular findings, treatment decision-making, and long-term outcomes in this rare patient population [5-7]. Here, we present the case of a 16-year-old patient with mucinous adenocarcinoma of the descending colon, deficient mismatch repair characterized by loss of MSH2/MSH6 expression, and a KRAS mutation. The immunohistochemical findings were highly suggestive of an underlying hereditary cancer syndrome and supported referral for genetic counseling and consideration of germline testing. This case highlights the importance of maintaining clinical suspicion for colorectal cancer in adolescents with atypical abdominal symptoms, integrating molecular diagnostics into routine clinical practice, and adopting a multidisciplinary approach to management.

## Case Report

A 16-year-old male initially underwent appendectomy for presumed acute appendicitis. Persistent postoperative bowel obstruction led to computed tomography (CT), which revealed a descending colon tumor requiring emergency Hartmann's procedure. Histopathology demonstrated mucinous adenocarcinoma, pT4N0M0, with vascular invasion. Immunohistochemistry showed loss of MSH2/MSH6 expression with preserved MLH1/PMS2, indicating dMMR and findings highly suggestive of





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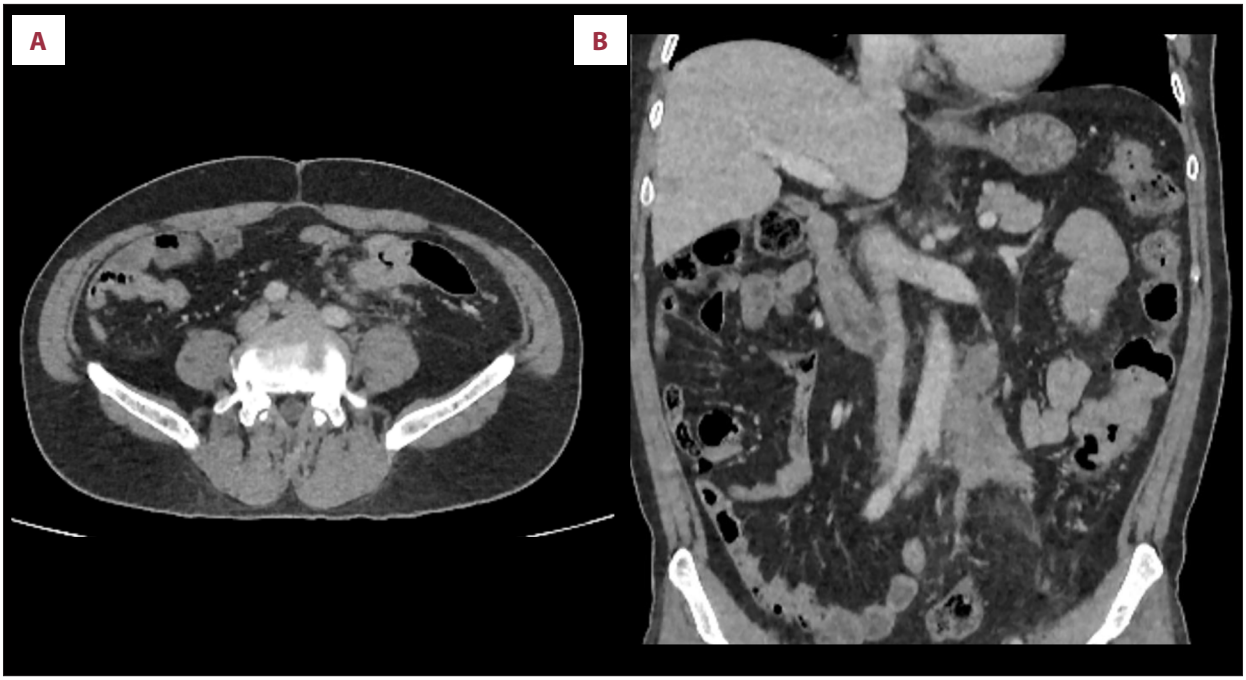


**Figure 1.** Contrast-enhanced abdominal computed tomography (CT). (**A**) Axial CT image demonstrating an irregular circumferential mass of the descending colon (arrow) causing marked luminal narrowing. (**B**) Coronal reconstruction showing eccentric thickening of the colonic wall with infiltration of the surrounding pericolic fat (arrowheads), consistent with locally advanced colorectal carcinoma and bowel obstruction.

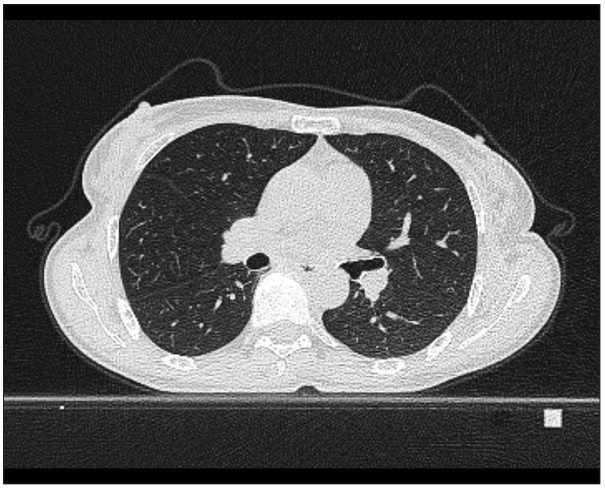

**Figure 2.** Contrast-enhanced chest computed tomography (CT). Axial chest CT demonstrating small bilateral pulmonary nodules (arrows) without radiological features suggestive of metastatic disease. These lesions remained stable throughout follow-up and showed no metabolic activity on PET/CT.

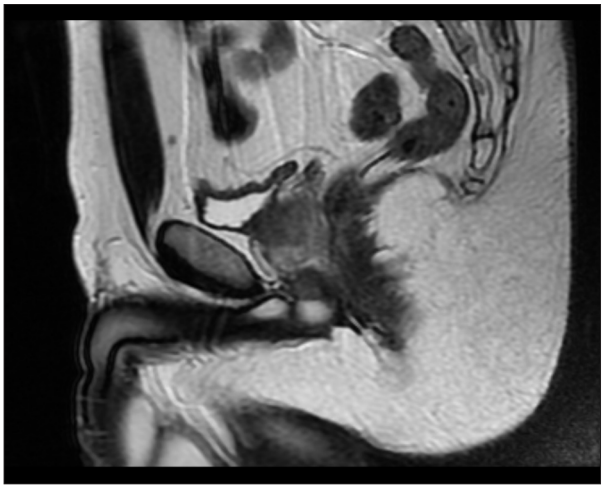

**Figure 3.** Pelvic magnetic resonance imaging (MRI). Pelvic MRI performed for staging demonstrated no evidence of pelvic tumor extension, enlarged pelvic lymph nodes, or metastatic disease.

Lynch syndrome, although germline testing was unavailable. Molecular testing identified a KRAS mutation without NRAS alterations. Because of multiple high-risk features, including T4 disease, bowel obstruction, vascular invasion, mucinous histology, and very young age, the patient received 12 cycles of adjuvant FOLFOX chemotherapy. More than 5 years after treatment, he remains disease-free.

This 16-year-old previously healthy male presented with progressive epigastric pain that subsequently migrated to the right lower quadrant and was accompanied by nausea, generalized weakness, and malaise. There was no reported family history







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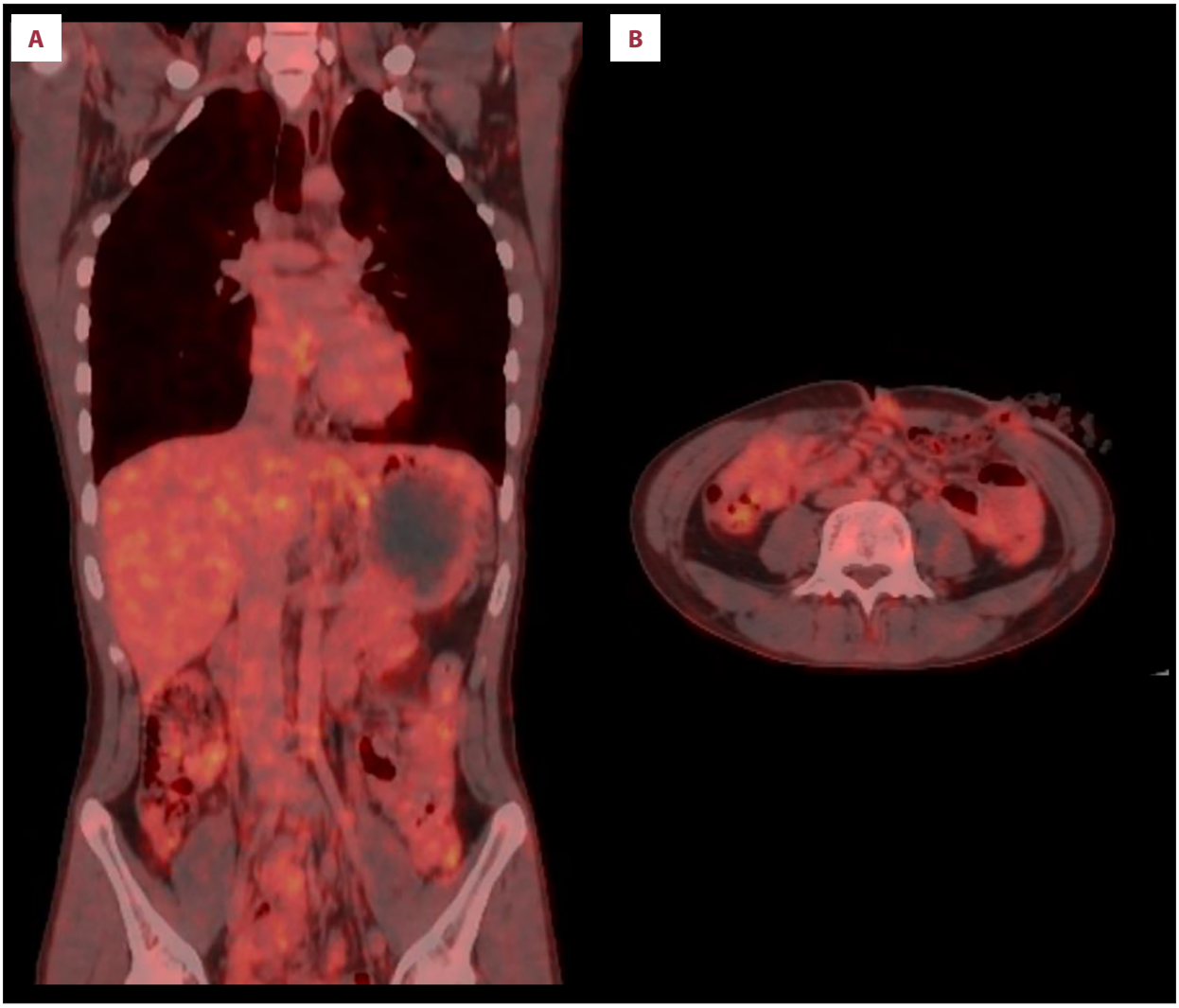


**Figure 4.** Positron emission tomography/computed tomography (PET/CT). (**A**) PET/CT demonstrating no abnormal hypermetabolic activity suggestive of distant metastatic disease. (**B**) Metabolically inactive left adrenal lesion (arrow), consistent with a benign adrenal adenoma.

of colorectal cancer or other Lynch syndrome-associated malignancies among first-degree relatives. Owing to worsening abdominal pain and clinical suspicion of acute appendicitis, the patient was admitted to the emergency surgical department and underwent appendectomy. Histopathological examination subsequently confirmed acute superficial appendicitis.

Approximately 2 days after surgery, he developed persistent abdominal pain, progressive abdominal distension, and clinical signs of early postoperative adhesive intestinal obstruction. Because of the atypical postoperative course, contrast-enhanced CT of the abdomen was performed. CT demonstrated an irregular circumferential mass measuring approximately 6.8 cm in the descending colon, associated with marked luminal narrowing and infiltration of the surrounding pericolic fat, suggesting transmural tumor extension (**Figure 1**). These findings raised strong suspicion for colorectal malignancy complicated by bowel obstruction.

The patient subsequently underwent emergency laparotomy with Hartmann's procedure and resection of the affected colonic segment. Histopathological examination revealed mucinous adenocarcinoma infiltrating through the entire bowel wall with serosal involvement. Additional pathological review demonstrated vascular tumor emboli, focal comedo-type necrosis, moderate peritumoral lymphocytic infiltration, and adjacent villous and serrated adenomas. Based on pathological findings, the tumor was classified as pT4N0M0 (stage IIB).

Postoperative staging investigations, including chest CT, pelvic magnetic resonance imaging (MRI), and positron emission tomography/computed tomography (PET/CT), demonstrated





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no evidence of locoregional recurrence or distant metastatic disease (**Figures 2-4**). Small bilateral pulmonary nodules were identified but remained below PET resolution and showed no metabolic activity during subsequent follow-up examinations. PET/CT also demonstrated a metabolically inactive left adrenal lesion consistent with a benign adrenal adenoma.

Comprehensive molecular and immunohistochemical analyses were performed approximately 2 months after surgery. Molecular testing identified a KRAS mutation, while NRAS mutations were not detected. Immunohistochemistry demonstrated complete loss of MSH2 and MSH6 protein expression with preserved MLH1 and PMS2 expression, confirming mismatch repair deficiency (dMMR). This immunophenotype is highly suggestive of an underlying hereditary mismatch repair defect and raised strong suspicion for Lynch syndrome; however, germline genetic confirmation was not available. Accordingly, the patient was considered to have suspected hereditary colorectal cancer and was recommended for genetic counseling and germline testing.

The patient's case was reviewed by a multidisciplinary tumor board. Although the benefit of adjuvant chemotherapy in stage II dMMR colon cancer remains controversial, the presence of several high-risk clinicopathological features—including T4 disease, bowel obstruction, vascular invasion, mucinous histology, and exceptionally young age at diagnosis—supported administration of adjuvant chemotherapy. The patient therefore received 12 cycles of the FOLFOX regimen consisting of oxaliplatin, leucovorin, and 5-fluorouracil. Treatment was completed without dose reductions or treatment delays. Toxicities were limited to grade 1-2 neutropenia, mild sensory peripheral neuropathy, and asthenia, all of which were managed conservatively.

Serial surveillance with CT and PET/CT performed during and after completion of chemotherapy demonstrated no evidence of local recurrence or distant metastatic disease (**Figures 5-9**). Approximately 1 year after completion of systemic therapy, intestinal continuity was successfully restored. At the most recent follow-up, more than 5 years after completion of treatment, the patient remained alive with no evidence of disease recurrence, representing a favorable long-term outcome despite the presence of multiple adverse pathological risk factors at initial diagnosis.



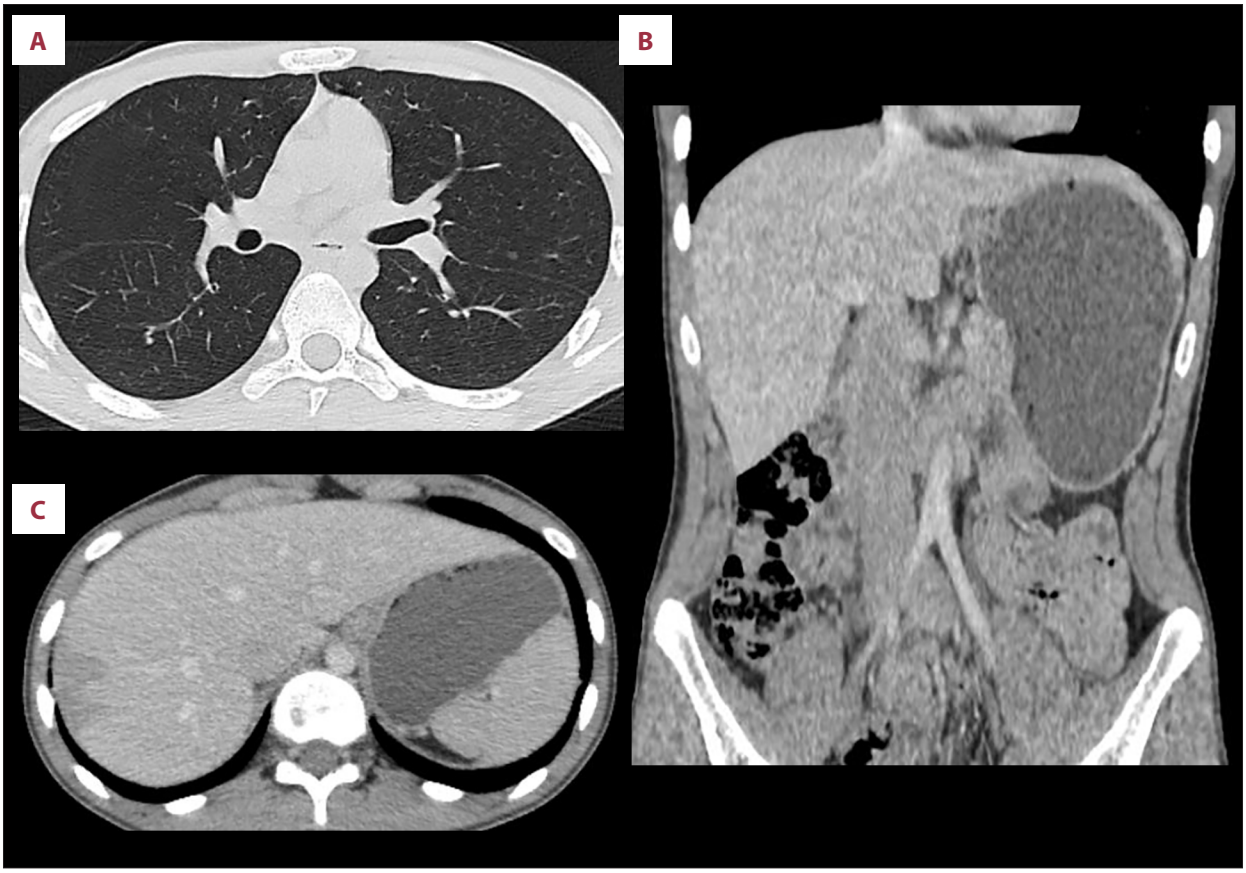


**Figure 5.** Follow-up contrast-enhanced computed tomography (CT) approximately 2 months after surgery.(**A-C**) Axial and coronal CT images demonstrating postoperative changes following Hartmann's procedure without evidence of local recurrence, lymph node enlargement, or distant metastatic disease. Previously identified pulmonary nodules remained unchanged.





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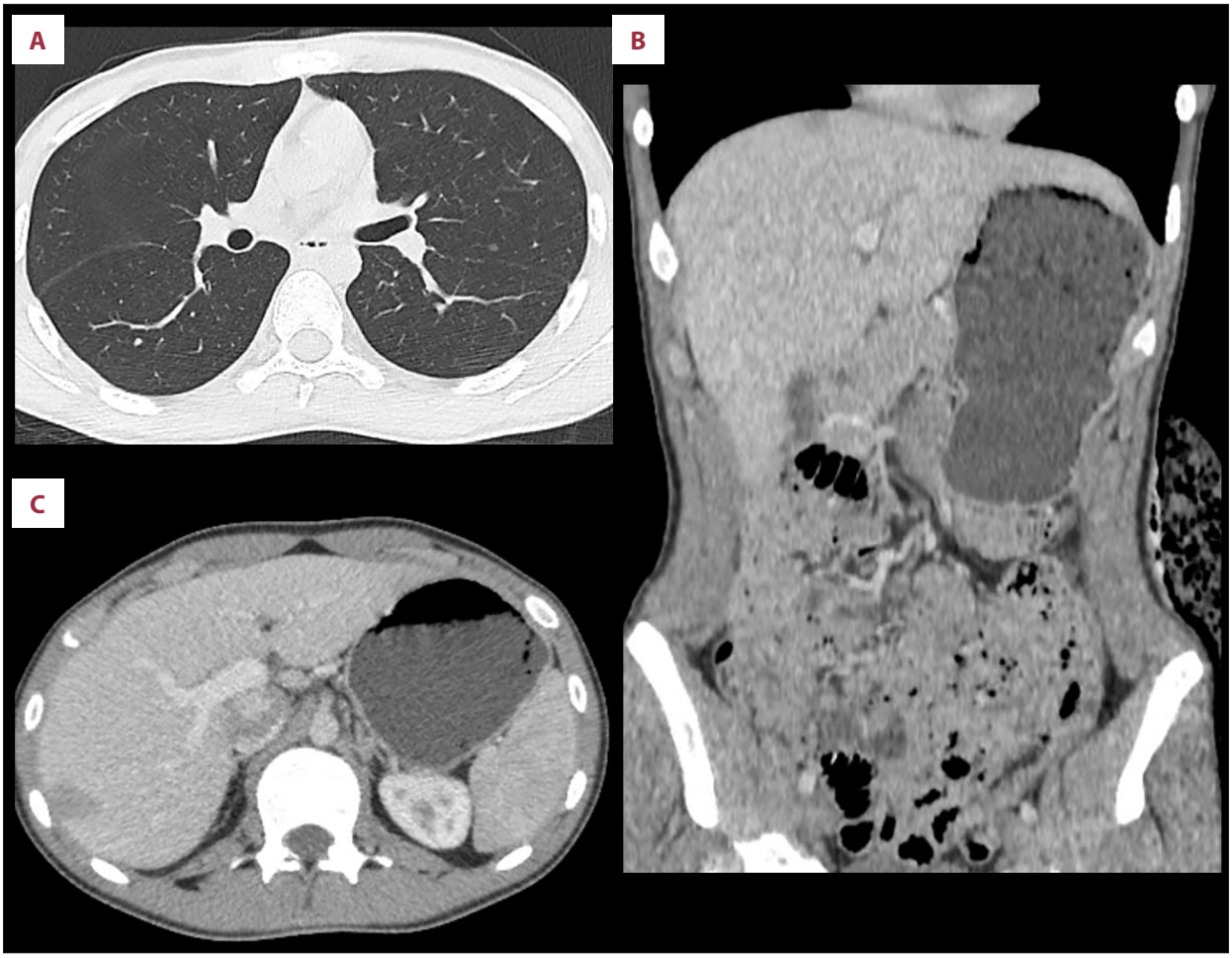


**Figure 6.** Follow-up contrast-enhanced computed tomography (CT) after completion of adjuvant chemotherapy. (**A-C**) CT images showing no radiological evidence of disease recurrence or metastatic progression. Postoperative findings remained stable, and no new lesions were identified.

## Discussion

This case illustrates several important diagnostic, pathological, and therapeutic aspects of colorectal cancer (CRC) occurring in adolescence. Although pediatric CRC is exceptionally rare, it is frequently associated with delayed diagnosis, advanced disease at presentation, and unfavorable histopathological features [5-7,23-25]. Because abdominal pain is a common symptom in children and adolescents, colorectal malignancy is rarely included in the initial differential diagnosis, often resulting in diagnostic delay [4-6,23]. In the present case, the patient's symptoms initially mimicked acute appendicitis, and the diagnosis of CRC became apparent only after persistent postoperative intestinal obstruction prompted further imaging. This clinical course emphasizes the importance of maintaining a broad differential diagnosis when young patients present with atypical or persistent abdominal symptoms [5,6,23].

Consistent with previous reports, the tumor demonstrated several pathological characteristics commonly observed in early-onset CRC, including mucinous histology, transmural invasion, vascular invasion, and locally advanced disease [5-7,24,25]. These features are generally associated with more aggressive tumor biology and poorer clinical outcomes than those observed in conventional adult-onset CRC [4-7]. Nevertheless, prognosis in young patients is influenced not only by tumor biology but also by timely diagnosis, complete surgical resection, and appropriate multidisciplinary management [16,17].

One of the most clinically relevant findings in this case was mismatch repair deficiency demonstrated by loss of MSH2 and MSH6 protein expression. This immunohistochemical pattern is highly suggestive of an underlying hereditary mismatch repair defect and appropriately raises suspicion for Lynch syndrome [12-15,22]. However, because germline genetic testing was not performed, a definitive diagnosis of Lynch syndrome could not be established on the basis of immunohistochemistry





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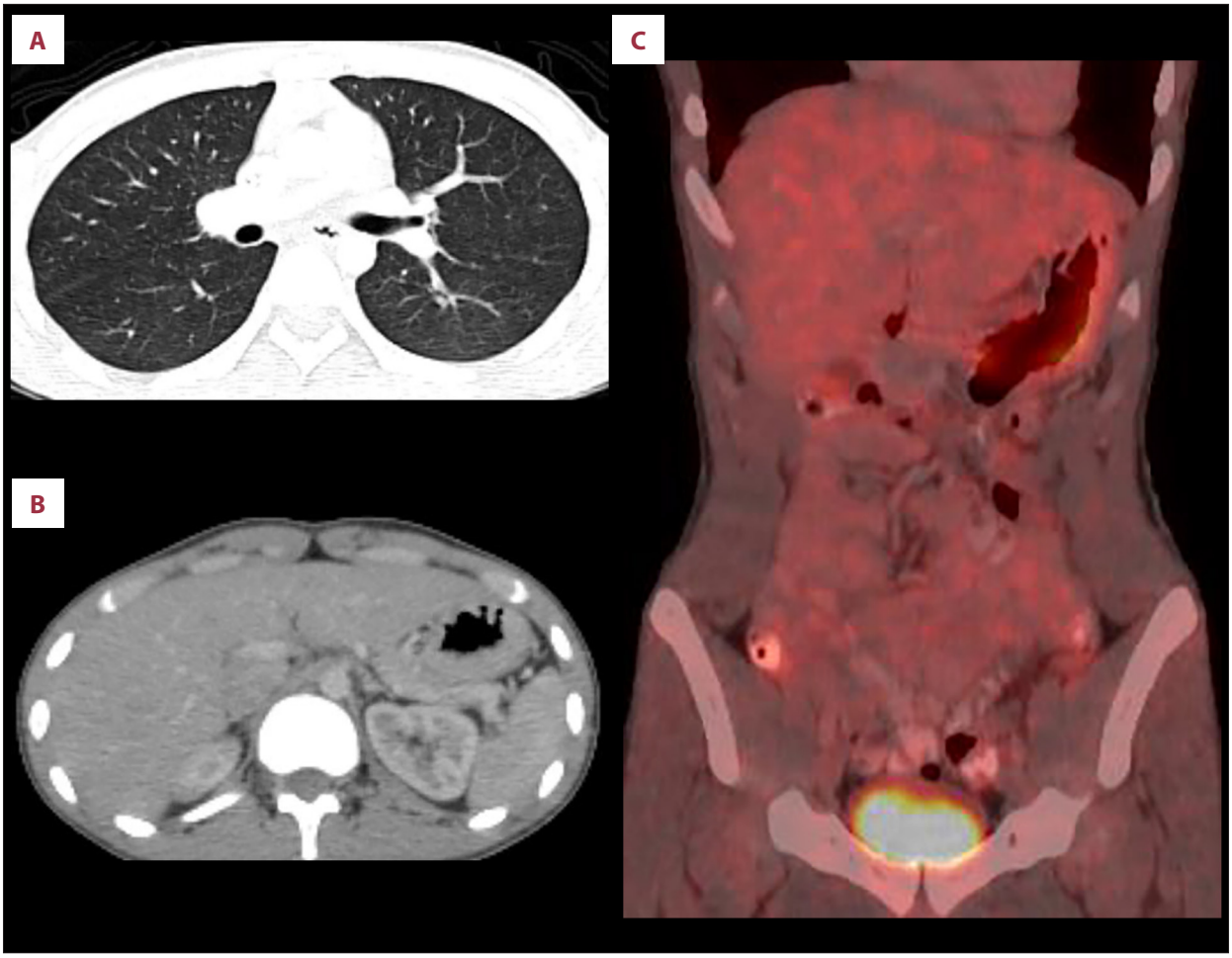


**Figure 7.** Follow-up computed tomography (CT) during surveillance. (**A-C**) CT examination demonstrating continued absence of local recurrence or distant metastases. Bilateral pulmonary nodules remained stable in size and appearance, consistent with benign lesions.

alone. Instead, these findings support referral for genetic counseling and germline testing in accordance with current ESMO and NCCN recommendations [13-16,22]. This distinction is important because confirmation of Lynch syndrome has implications not only for the affected patient but also for surveillance and risk assessment among family members [13,15,22].

The coexistence of dMMR and a KRAS mutation is an additional molecular feature of interest. KRAS mutations occur in approximately 35% to 45% of colorectal cancers overall and may also be observed in tumors with deficient mismatch repair [19,30-33]. Nevertheless, KRAS mutations should be interpreted as somatic driver alterations rather than evidence confirming or excluding an inherited cancer syndrome [30-33]. In the present case, identification of the KRAS mutation did not influence initial treatment but may become clinically relevant if systemic therapy is required in the event of future disease recurrence [31,32].

Selection of postoperative treatment required careful multidisciplinary discussion. Current international guidelines indicate that patients with stage II dMMR colon cancer generally derive limited benefit from fluoropyrimidine monotherapy, whereas oxaliplatin-containing adjuvant chemotherapy may be considered in selected patients with high-risk pathological features [14,16,17,34]. In this patient, the combination of T4 disease, bowel obstruction, vascular invasion, mucinous histology, and exceptionally young age supported the decision to administer adjuvant FOLFOX chemotherapy. Although the favorable long-term outcome observed in this patient cannot be attributed to any single therapeutic intervention, it suggests that individualized management based on clinicopathological and molecular characteristics can be appropriate in selected high-risk patients [16,17,34].

The prolonged disease-free survival observed during follow-up further highlights the importance of comprehensive multidisciplinary care, including accurate pathological assessment,







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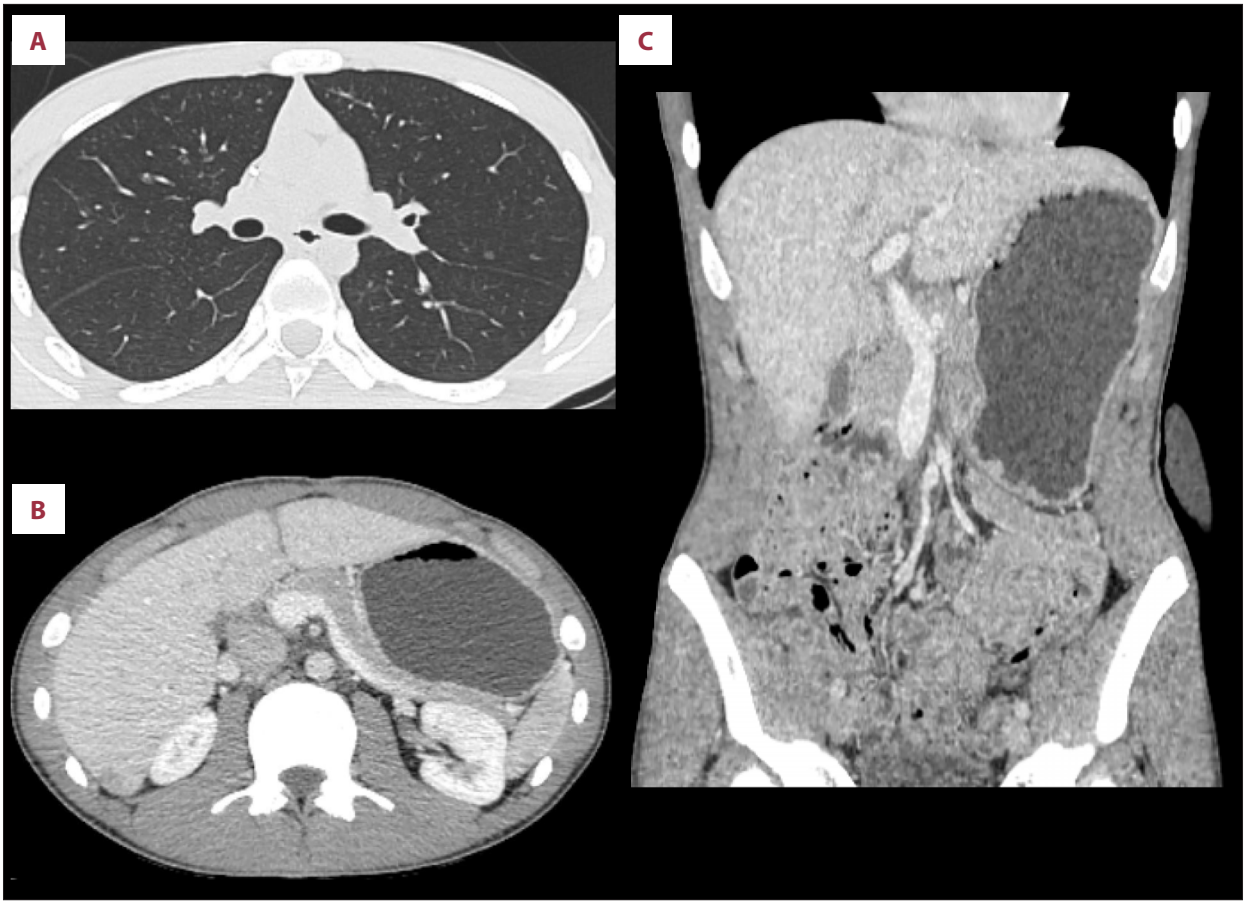


**Figure 8.** Surveillance imaging after restoration of intestinal continuity. (**A-C**) Follow-up computed tomography images demonstrating normal postoperative appearance after restoration of bowel continuity, without evidence of recurrent tumor, lymphadenopathy, or metastatic disease.

molecular characterization, radical surgery, carefully selected adjuvant therapy, and structured surveillance [14,16,17]. However, because this report describes a single patient, no conclusions regarding the superiority of a particular treatment strategy can be drawn. Larger multicenter studies and prospective registries are needed to better define optimal management strategies for adolescents with dMMR colorectal cancer [4,5,16].

Despite its limitations, this case provides several practical messages. First, colorectal cancer, although rare, should be considered in adolescents presenting with persistent or atypical abdominal symptoms [5-7]. Second, universal assessment of mismatch repair status is essential in young-onset CRC because it identifies patients who may require genetic counseling and germline evaluation for hereditary cancer syndromes [12-16]. Finally, integration of clinical, pathological, and molecular findings within a multidisciplinary framework may facilitate individualized clinical decision-making in complex cases such as the one presented here [16,17,22].

## Conclusions

This case emphasizes that colorectal cancer should be considered in adolescents with persistent or atypical abdominal symptoms. Routine MMR/MSI testing and comprehensive molecular profiling are essential in young-onset CRC to identify patients who may require genetic counseling and tailored management. Although limited to a single case, this report highlights the value of multidisciplinary decision-making and individualized treatment based on clinicopathological and molecular characteristics.

This case illustrates the diagnostic and therapeutic challenges associated with colorectal cancer in adolescents. The combination of mucinous histology, locally advanced disease, mismatch repair deficiency, and a KRAS mutation highlights the importance of comprehensive pathological and molecular evaluation in young patients with colorectal cancer. In this patient, loss of MSH2 and MSH6 expression on immunohistochemistry was highly suggestive of an underlying hereditary mismatch repair defect and supported referral for genetic counseling and





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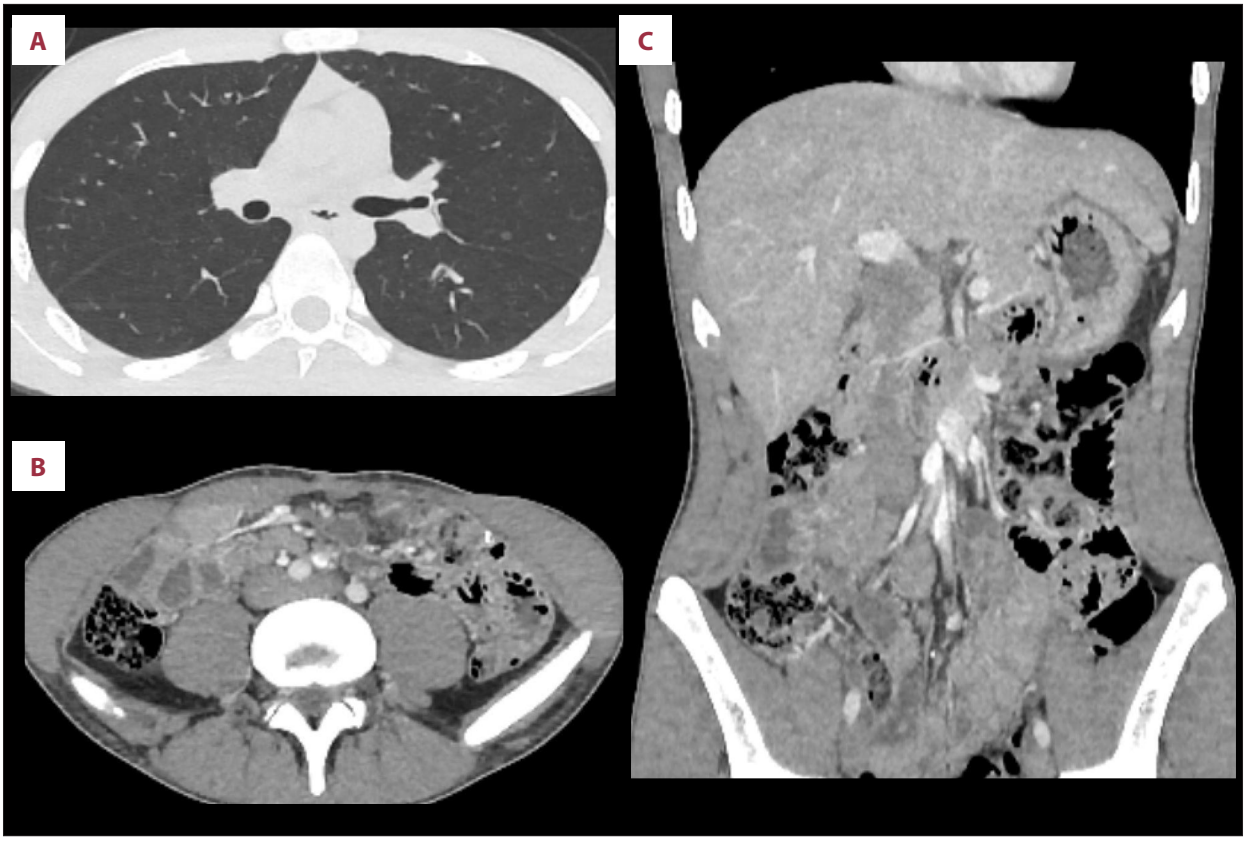


**Figure 9.** Long-term follow-up computed tomography (CT). (**A-C**) CT examination performed more than 5 years after initial treatment demonstrating persistent complete remission, with no evidence of local recurrence, distant metastases, or radiological progression.

consideration of germline testing, although Lynch syndrome could not be confirmed without genetic analysis.

Management was guided by a multidisciplinary approach that integrated clinicopathological risk factors with molecular findings. Although the role of adjuvant chemotherapy in stage II dMMR colorectal cancer remains controversial, the presence of multiple high-risk pathological features supported the use of postoperative FOLFOX chemotherapy in this individual patient. The favorable long-term disease-free survival observed during follow-up demonstrates that individualized treatment strategies can achieve excellent clinical outcomes in selected high-risk cases.

As a single case report, this study cannot establish treatment recommendations or determine the prognostic significance of specific molecular alterations. Nevertheless, it emphasizes the importance of maintaining clinical suspicion for colorectal cancer in adolescents presenting with persistent or atypical abdominal symptoms, performing routine MMR/MSI testing in young-onset colorectal cancer, and integrating clinical, pathological, and molecular findings into individualized patient management. Multicenter studies are needed to better define optimal diagnostic and therapeutic approaches for adolescents with colorectal cancer.

### Department and Institution Where Work Was Done

Department of Oncology named after Professor S.N. Nugmanov, Kazakh Research Institute of Oncology and Radiology, Almaty, Kazakhstan.

### Patient Consent Statement

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

### Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.







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