International Journal of Community Medicine and Public Health
Suliman O et al. Int J Community Med Public Health. 2026 Jun;13(6):3118-3121
http://www.ijcmph.com
pISSN 2394-6032 | eISSN 2394-6040

DOI: https://dx.doi.org/10.18203/2394-6040.ijcmph20261805

Case Report

Anorectal mucinous adenocarcinoma in a child: a case report

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Received: 31 January 2026
Accepted: 26 March 2026

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ABSTRACT

Primary gastrointestinal malignancies constitute approximately 2% of pediatric neoplasms, and colorectal carcinoma is the second most common gastrointestinal malignancy after primary liver tumors. Anorectal adenocarcinoma in children is exceptionally rare, accounting for less than 1% of all colorectal cancers and is often diagnosed at an advanced stage due to nonspecific symptoms. We describe the case of a 12-year-old boy who presented with a 3-month history of recurrent abdominal pain, diarrhea mixed with mucus and blood, and intermittent rectal bleeding, and was found to have a rectal mass. A per-rectal biopsy confirmed the diagnosis of adenocarcinoma. Computed tomography of the abdomen and pelvis revealed locally advanced disease without distant metastasis. The patient was evaluated by a multidisciplinary team to determine the optimal therapeutic approach. Anorectal adenocarcinoma in children remains a rare but aggressive malignancy with a poor prognosis if diagnosis is delayed. Early recognition, prompt biopsy of suspicious lesions, comprehensive staging, and genetic evaluation are essential for optimal management. Surgical resection remains the mainstay of treatment, often combined with neoadjuvant and adjuvant therapies. Reporting such rare cases contributes to improved awareness and may aid in developing evidence-based management strategies.

Keywords: Anorectal mucinous adenocarcinoma, Gastrointestinal malignancies, Delayed diagnosis

INTRODUCTION

Anorectal adenocarcinoma in the pediatric population is a rare but highly aggressive malignant neoplasm characterized by distinct clinicopathological and biological features.[1] Although colorectal carcinoma is common in adults, its occurrence in children remains exceptionally uncommon worldwide, with an estimated incidence of approximately one case per million.[2] Because of this rarity, the diagnosis is often not initially considered, leading to frequent delays in recognition and treatment.[3]

Most pediatric cases are diagnosed during adolescence, whereas occurrence in younger children is exceedingly rare.[4] Pediatric anorectal tumors tend to demonstrate more aggressive biological behavior compared with adult counterparts.[5] Mucinous adenocarcinoma represents the most prevalent histological subtype, followed by signet-ring cell carcinoma, both of which are associated with poor differentiation, rapid local invasion, and adverse outcomes.[6]

Delayed diagnosis contributes significantly to poor prognosis in this population.[7] Early clinical manifestations are subtle and nonspecific, most commonly presenting as rectal bleeding, altered bowel habits, constipation, abdominal pain, or tenesmus.[2] These symptoms frequently mimic benign anorectal disorders

such as anal fissures or hemorrhoids, leading to delayed referral and advanced presentation.[8]

A comprehensive diagnostic evaluation is essential. Endoscopic examination with biopsy remains the gold standard for tissue diagnosis, while cross-sectional imaging such as MRI or CT is necessary for accurate staging and surgical planning.[5] Histopathological confirmation helps differentiate these malignancies from other childhood rectal lesions, such as lymphoma, rhabdomyosarcoma, or inflammatory polyps.[4]

Genetic predisposition must also be considered, as several pediatric cases have been associated with hereditary cancer syndromes, including familial adenomatous polyposis (FAP), Lynch syndrome, and hereditary nonpolyposis colorectal cancer (HNPCC).[9] Early molecular evaluation allows for the identification of at-risk family members and the personalization of surveillance strategies.[10]

Recent advances in molecular oncology suggest that pediatric anorectal adenocarcinomas may harbor unique genetic and epigenetic alterations distinct from those seen in adult forms.[3] Understanding these molecular signatures could lead to more effective targeted therapies and contribute to improved future outcomes.[11]

Multidisciplinary management involving pediatric surgeons, oncologists, radiologists, and genetic counselors is vital for optimal outcomes.[12] Surgical resection remains the cornerstone of therapy, with chemotherapy and radiotherapy serving as important adjuncts, particularly in advanced disease.[13] Ultimately, greater awareness among pediatricians and surgeons, combined with prompt investigation of persistent anorectal symptoms, is crucial for earlier diagnosis and better survival outcomes in affected children.[7]

CASE REPORT

A 12-year-old boy presented with a 3-month history of recurrent abdominal pain, diarrhea mixed with mucus and blood, and persistent rectal bleeding. He also reported fever, anorexia, progressive weight loss, and occasional episodes of abdominal distention and vomiting. Subsequently, he developed absolute constipation, worsening abdominal distention, and bilious vomiting. He was admitted to a local hospital, where a diagnosis of acute intestinal obstruction was made. Emergency laparotomy revealed a rectal tumor, for which a diverting loop transverse colostomy was performed.

Following initial management, the patient was referred to the national center for pediatric surgery for further evaluation. Upon admission, he appeared ill, cachectic, but was hemodynamically stable. Abdominal examination showed a healed midline laparotomy scar and a functioning right-sided loop transverse colostomy. A visible bulge was noted in the suprapubic and infra-umbilical regions, which on palpation was found to be a firm, nodular intra-abdominal mass. Digital rectal examination revealed blood-stained mucus discharge and a markedly reduced sphincter tone. Approximately 2 cm from the anal verge, a fixed circumferential mass completely obliterating the rectal lumen was palpated. Shown in Figure 1: EUA view of rectal tumor.

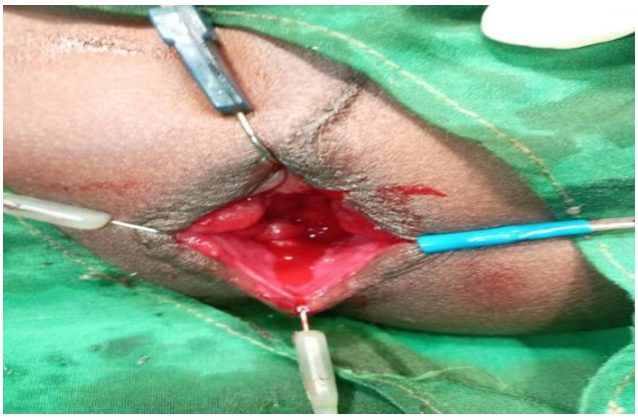

Figure 1: EUA view of rectal tumor.

An incisional per-rectal biopsy from the mass demonstrated ill-formed, fragmented glandular structures and abundant mucin pools invading the stroma and adjacent normal mucosa, consistent with grade 1 invasive mucin-secreting rectal adenocarcinoma. Shown in Figure 2 and 3: Histopathology slide.

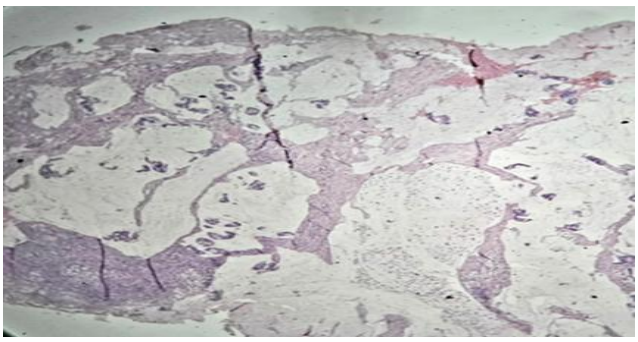

Figure 2: Histopathology slide.

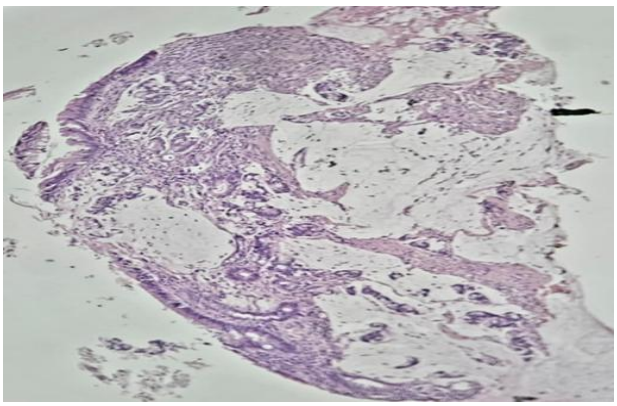

Figure 3: Histopathology slide.

CT scan of the abdomen and pelvis revealed significant mural thickening with heterogeneous enhancement, multiple focal calcifications, and luminal obliteration of the rectum and anal canal, shown in Figure 4: CT cuts. Multiple enlarged lymph nodes were identified in the perirectal fat and along the bilateral internal iliac chains, corresponding to Dukes' C stage locally advanced rectal cancer.

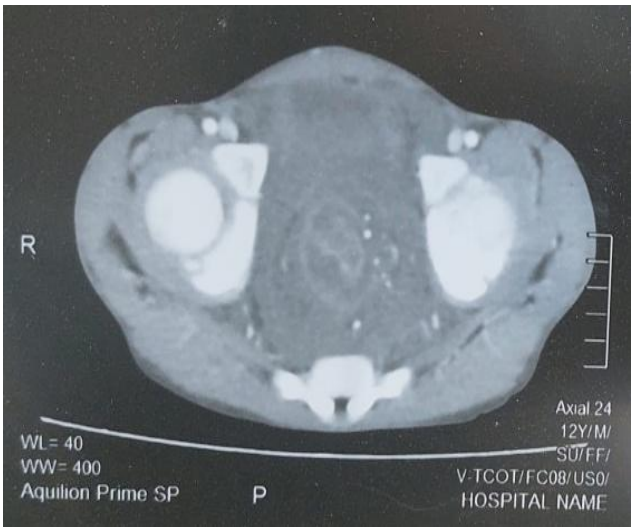

Figure 4: CT cuts.

Baseline laboratory investigations showed anemia, elevated inflammatory markers, and hypoalbuminemia, consistent with chronic disease and malnutrition. Chest CT and liver ultrasound were obtained to rule out distant metastases and showed no secondary deposits. Differential diagnoses at the time of presentation included rectal lymphoma, inflammatory bowel disease, and chronic infective colitis, which were excluded by histopathological confirmation. A multidisciplinary tumor board recommended neoadjuvant chemoradiation therapy followed by surgical resection and possible adjuvant chemotherapy. However, after counseling the parents about the malignant nature of the disease and the need for multimodal therapy, they refused further management, and the child was discharged against medical advice.

DISCUSSION

Anorectal adenocarcinoma in the pediatric population is exceedingly rare, with an estimated incidence of approximately 1 case per million children.[1] Most reported cases occur in patients older than 10 years, and no consistent gender predilection has been observed. Common presenting symptoms include rectal bleeding, altered bowel habits, abdominal pain, tenesmus, and weight loss.[2] Because these features overlap with benign anorectal disorders such as fissures or hemorrhoids, diagnosis is often delayed until the disease is locally advanced or metastatic at presentation.[3]

Histologically, pediatric anorectal adenocarcinoma displays distinct characteristics compared to adult-onset disease.[4] Mucinous differentiation occurs in 50–60% of cases, while signet-ring cell morphology is seen in about 20%, both subtypes being correlated with poor differentiation, extensive local spread, and unfavorable prognosis.[5] In contrast to adult cases, which are often sporadic, several pediatric occurrences have been linked to underlying genetic predispositions or hereditary syndromes.[9] Recent molecular studies have documented frequent mismatch repair (MMR) deficiency and microsatellite instability (MSI-high) status in this group, underscoring the importance of molecular testing.[3] Mutations in KRAS, BRAF, and TP53 genes have also been observed, although they occur less commonly than in adults.[10]

A high index of clinical suspicion is essential for timely diagnosis.[7] Digital rectal examination remains the most accessible and critical initial step and should not be overlooked in children with persistent rectal bleeding or altered bowel habits.[4] Endoscopic evaluation with biopsy is mandatory for histopathological confirmation.[5] Magnetic resonance imaging (MRI) or computed tomography (CT) of the pelvis assists in accurate local staging, assessing tumor depth, and evaluating mesorectal fascia involvement.[5] Whole-body CT or PET-CT scans are recommended to identify lymphatic or hematogenous metastases.[12] In addition, every confirmed case should undergo MMR and MSI testing to guide both treatment options and family screening for hereditary cancer syndromes.[3]

Management strategies for pediatric anorectal adenocarcinoma largely mirror adult treatment protocols but must consider growth potential, psychosocial aspects, and long-term survivorship.[13] Radical surgical resection with negative margins (R0 resection) remains the cornerstone of curative treatment.[12] Depending on tumor location and sphincter involvement, procedures such as low anterior resection (LAR) or abdominoperineal resection (APR) are employed.[12] In suitable patients, neoadjuvant chemoradiation can downstage tumors and increase sphincter preservation rates.[13] Chemotherapy regimens such as FOLFOX or CAPOX are typically used, though tolerance and toxicity profiles must be closely monitored in children.[13] The use of radiation therapy is limited by the potential for growth impairment, bowel dysfunction, and infertility, requiring careful individualized planning.[10]

Recent therapeutic advances include immunotherapy, particularly PD-1 inhibitors such as pembrolizumab and nivolumab, which demonstrate encouraging responses in MSI-high/MMR-deficient tumors.[3] However, clinical experience in pediatric populations remains limited, and long-term outcomes are yet to be established.[11] Genetic counseling should be systematically offered to affected families, as identifying germline mutations can facilitate early detection in at-risk relatives.[9]

Despite aggressive multimodal therapy, pediatric anorectal adenocarcinoma carries a dismal prognosis compared with adult colorectal malignancy.[1] The overall five-year survival rate generally remains below 30%, largely due to delayed diagnosis and advanced disease stage at presentation.[7] Early detection, complete surgical resection, and favorable molecular profiles such as MSI-high status are among the few prognostic indicators associated with improved survival.[3] Lifelong surveillance is essential, given the risk of recurrence and potential late effects of therapy, emphasizing the need for structured follow-up protocols.[14] Increasing awareness among pediatricians and surgeons about this rare entity is therefore critical to reduce diagnostic delays and enhance patient outcomes.[7]

CONCLUSION

The rarity of anorectal adenocarcinoma in children presents significant diagnostic and therapeutic challenges. The predominance of mucinous and signet-ring cell subtypes contributes to its aggressive biological behavior and poor overall prognosis.

Early clinical recognition, prompt biopsy of suspicious anorectal lesions, comprehensive radiological staging, and detailed genetic evaluation are crucial for accurate diagnosis and optimal treatment planning. Surgical resection remains the cornerstone of therapy, while adjuvant chemotherapy and radiotherapy should be tailored to the stage, molecular profile, as well as treatment tolerance of the individual patient.

Although management principles are largely extrapolated from adult colorectal cancer protocols, treatment strategies must be adapted to account for pediatric anatomy, ongoing growth, and psychosocial development. Incorporating molecular profiling into the therapeutic algorithm allows for more precise, personalized care, particularly in MMR-deficient or MSI-high tumors that may respond to immunotherapy.

Long-term multidisciplinary follow-up is vital to monitor recurrence, manage functional outcomes, and address late treatment-related morbidity. Increasing awareness among pediatricians and surgeons is essential to ensure early referral and reduce diagnostic delays.

Future research should prioritize collaborative multicenter studies and population-based cancer registries to define evidence-based, pediatric-specific management guidelines. The integration of genomic insights and registry data will ultimately help refine risk stratification, improve survival outcomes, and enhance quality of life for affected children.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Suliman O, Arbab Z, Babiker S, Basheir A, Nugud F. Anorectal mucinous adenocarcinoma in a child: a case report. Int J Community Med Public Health 2026;13:3118-21.