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Case report

Gastric Adenocarcinoma in Two Adolescents with Genetic Predisposition: A Case Report

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Abstract

Introduction: Gastric tumors in pediatric patients are extremely rare, posing diagnostic and therapeutic challenges due to limited medical literature and nonspecific clinical presentation. **Case report:** Two cases of 13-year-old adolescents with no relevant medical history are reported, both presenting with nonspecific gastrointestinal symptoms. Both were diagnosed with stage IV diffuse gastric adenocarcinoma through upper gastrointestinal endoscopy with biopsy and immunohistochemistry. Rapid tumor progression and resistance to chemotherapy were observed. The first case was associated with Lynch syndrome, and the second met criteria for hereditary diffuse gastric cancer (HDGC). **Discussion:** These tumors exhibit high aggressiveness and poor response to treatment, which is associated with delayed diagnosis and probable genetic predisposition. Currently, no specific pediatric protocols exist; therefore, management is based on adult guidelines, with limited outcomes in this population. **Conclusion:** It is essential to maintain a high index of clinical suspicion in the presence of persistent or nonspecific symptoms, implement early genetic screening, and adopt a multidisciplinary approach. Further research on hereditary syndromes and the development of pediatric-specific guidelines are needed to achieve earlier diagnosis and improve prognosis.

Keywords

Gastric cancer; adenocarcinoma; adolescent; pediatrics; abdominal pain.

INTRODUCTION

Gastrointestinal malignant tumors commonly occur during the sixth and seventh decades of life, whereas in childhood and adolescence they are extremely rare and aggressive, accounting for approximately 5% of all pediatric neoplasms. Gastric adenocarcinoma represents 0.05% of all gastrointestinal neoplasms[1]. Gastric carcinoma exhibits aggressive behavior and is usually diagnosed at advanced stages because its initial symptoms are nonspecific, including abdominal pain, emesis, anemia, diarrhea, and weight loss[2,3].

In children and adolescents, the genetic factor becomes more relevant and is usually present in 10% of cases[4]. Clinical aggressiveness may be associated with hereditary syndromes such as hereditary diffuse gastric cancer (HDGC) caused by a mutation in the CDH1 gene, or Lynch syndrome due to loss of expression of MLH1, MSH2, MSH6, or PMS2. Both exhibit an autosomal dominant inheritance pattern and extensive metastatic dissemination. The diffuse subtype, characterized by the presence of signet ring cells, confers a poorer prognosis and poor response to chemotherapy, sometimes associated with a family history of cancer or occurring in its absence[5,6].

In Lynch syndrome, cancer may develop in different areas of the gastrointestinal tract, including the stomach, small intestine, and pancreas, although in most cases the

cancer is detected in the colon[7]. Isolated breast cancer is not typically associated with pathogenic variants in the MSH6 gene; however, such cases have been reported[8].

Although *Helicobacter pylori* infection and certain lifestyle habits have been recognized as relevant risk factors for the development of gastric cancer in adults, this association is less clear in the pediatric population. Nevertheless, in regions with a high prevalence of *H. pylori*, a hereditary mutation in the E-cadherin gene has been identified, suggesting a possible genetic and environmental relationship in the early onset of this disease[1,9]. In Colombia, 50% of middle-class children and nearly 80% of children from low socioeconomic backgrounds are infected with *H. pylori* before 10 years of age, and the infection is associated with positive cagA, vacA, and iceA genes[10].

The genetics of gastric cancer reveal significant heterogeneity with distinct molecular subtypes, including Epstein-Barr virus (EBV)-positive tumors, microsatellite instability (MSI), chromosomal instability (CIN), and genomically stable (GS) tumors[11]. This highlights the importance of considering molecular classification together with the evaluation of hereditary syndromes in adolescents with gastric cancer in order to establish targeted therapies and avoid unfavorable outcomes. For these reasons, we present the report of two cases of adolescents with metastatic diffuse gastric adenocarcinoma in whom molecular alterations associated with hereditary genetic predisposition were identified.

CASE REPORT

Case 1

This was a previously healthy 13-year-old female patient with a maternal history of breast cancer. She was referred to a tertiary care center in Cali, Colombia, due to a clinical presentation of severe persistent osteomuscular pain in the dorsolumbar region of approximately 10 days' duration, which had been associated with sports-related trauma, accompanied by mucocutaneous pallor and one episode of mild hematemesis. The patient also reported diffuse moderate-intensity abdominal pain, loss of appetite, and nonspecific weight loss. On admission physical examination, generalized mucocutaneous pallor, abdominal tenderness on palpation, and tachycardia were observed. She subsequently experienced syncope and was hospitalized.

Admission laboratory tests showed severe anemia with a hemoglobin level of 5.0 g/dL, requiring blood product transfusion. Given the suspicion of gastrointestinal bleeding, an upper gastrointestinal endoscopy (UGE) was performed, revealing an extensive exophytic lesion involving the lesser curvature up to the antropyloric junction, with multiple sites of erosion and scant layered bleeding, along with a report of chronic superficial gastritis and negative *H. pylori* testing. A myeloproliferative syndrome was initially suspected; therefore, a bone marrow biopsy was performed, in which no cellular populations suspicious for neoplasia were identified. Infectious diseases were ruled out.

Imaging studies, including magnetic resonance imaging of the abdomen and chest, demonstrated a nodal conglomerate involving multiple thoracic and lumbar vertebral bodies, as well as the iliac wings. Consequently, the patient underwent greater omentum biopsy by pediatric surgery because of suspicion of gastric lymphoma. Histopathological examination revealed poorly differentiated diffuse gastric adenocarcinoma with signet ring cells. Immunohistochemistry (IHC) showed negative microsatellite instability and programmed death-ligand 1 (PD-L1), negative fluorescence *in situ* hybridization (FISH) for human epidermal growth factor receptor 2 (HER2), and loss of nuclear expression of mutS homolog 2 (MSH2), findings suggestive of Lynch syndrome.

Subsequently, during hospitalization, the patient developed bilateral breast masses. Given the family history and suspicion of Lynch syndrome, breast ultrasonography was performed, reporting bilateral BI-RADS 5 solid focal lesions, with pathology compatible with metastatic adenocarcinoma, possibly originating from the upper gastrointestinal tract as the primary site. The patient was classified as stage IV disease, and palliative chemotherapy with the FLOT regimen (fluorouracil, leucovorin, oxaliplatin, and docetaxel) was initiated (**Table 1**). However, disease progression occurred during treatment; palliative sedation was started, and the patient died two months after diagnosis.

Case 2

A previously healthy 13-year-old male patient, with no relevant personal history or reported family history of cancer, was referred to a tertiary care center in Cali, Colombia, because of severe colicky abdominal pain of one and a half months' duration. On admission physical examination, he appeared with mild mucocutaneous pallor, adynamia, mesogastric abdominal pain, and tachycardia, without palpable abdominal masses or lymphadenopathy; therefore, he was hospitalized. Subsequently, the patient developed hematemesis and melena. He required blood product transfusion because of severe anemia with a hemoglobin level of 6.4 g/dL.

An upper gastrointestinal endoscopy (UGIE) was performed, reporting an extensive excavated gastric ulcer measuring 2.5 to 3 cm in diameter, located on the greater curvature at the antropyloric junction, with raised borders and positive *H. pylori* testing, which was treated with triple

therapy. Complementary tests were performed, including a negative infectious profile and diagnostic imaging with abdominal computed tomography (CT), which showed gastric wall thickening with lymphadenopathy adjacent to the celiac trunk and scant free fluid in the pelvic cavity. In addition, positron emission tomography (PET scan) demonstrated metastatic involvement of the bone marrow. Pediatric hematology-oncology requested bilateral bone marrow aspiration, which was negative for malignancy, as well as immunohistochemistry (IHC) to confirm the diagnosis, reporting HER2 negativity and a pathogenic variant in CDH1. Variants of uncertain significance in MSH2 and MUTYH were also identified.

The patient was diagnosed with stage IV hereditary diffuse gastric cancer (HDGC), and neoadjuvant chemotherapy with 5-fluorouracil and cisplatin was initiated for three cycles (**Table 1**). Surgical local tumor resection was considered; however, the patient showed stable disease progression, and a repeat PET scan demonstrated signs of tumor progression in the bone marrow, with persistence of nodal and gastric disease. The patient was considered chemotherapy-resistant, subsequently received palliative radiotherapy, and died nine months after diagnosis.

Table 1. Stage-Based Treatment of Pediatric Gastric Cancer

Stage	Treatment
I	Radical gastrectomy with lymphadenectomy. Adjuvant chemotherapy may be considered in high-risk cases.
II	Radical surgery if resectable. Neoadjuvant chemotherapy ± radiotherapy according to tolerance.
III	If resectable: surgery + neoadjuvant chemotherapy. If unresectable: systemic chemotherapy. Radiotherapy may be added as palliative or neoadjuvant treatment.
IV	Palliative treatment: supportive surgery (obstruction, bleeding), palliative chemotherapy, and radiotherapy. Clinical trials are strongly recommended. Comprehensive management in specialized pediatric oncology centers.

Author's own research.

DISCUSSION

Primary gastric tumors in children are uncommon, and gastric carcinoma is even more unusual. Its prognosis depends on the extent of disease at the time of diagnosis and the appropriateness of treatment, which has resulted in limited available information regarding this pathology in the pediatric population. Nevertheless, one of the largest studies conducted was performed by Tessler and colleagues[3] in 2019 using records from the National Cancer Data Base between 2004 and 2014. Their objective was to compare the clinical presentation, management, and outcomes between pediatric patients (≤21 years) and adults (>21 years) with gastric adenocarcinoma. They identified that, among the 129,024 total cases, only 129 cases (0.10%) corresponded to pediatric patients. In addition, these patients more frequently presented with advanced-stage disease (stage IV) (56%) compared with adults (41%). They concluded that although children had more advanced disease and more aggressive histological types, their survival rates were similar to those of adults.

Likewise, reports of this disease in the pediatric population are rare in Colombia[15]. However, two case reports from 2007 and 2021 were identified. The first involved a 12-year-old male adolescent from Popayán, Cauca, who presented with a one-year history of diffuse symptoms (abdominal pain, vomiting, melena, hematemesis), and biopsy revealed diffuse-type adenocarcinoma with positive *H. pylori* immunohistochemistry and the presence of cagA and vacA genes. He underwent total gastrectomy, and metastases to the spleen, pancreas, and colon were identified[12]. The second case involved a 10-year-old girl from Medellín, Antioquia, who presented with constitutional syndrome (abdominal pain, postprandial emesis, and weight loss); studies revealed poorly differentiated diffuse gastric adenocarcinoma with signet ring cells, and diagnostic laparoscopy showed signs of peritoneal carcinomatosis, for which palliative chemotherapy was offered[13].

As occurred in the previously described cases, our patients presented with metastatic disease from the time of diagnosis and showed resistance to chemotherapy, demonstrating more aggressive tumor behavior than that observed in the adult population[3]. This underscores the need to implement interventions that enable early detection of gastric cancer in this population because, as reported in the literature and also observed in these patients, the clinical manifestations of this type of cancer are highly nonspecific in the early stages. More striking symptoms, such as severe epigastric abdominal pain, weight loss, and hematemesis, are observed in advanced stages, which is generally when the disease is diagnosed[14].

Another exceptionally rare gastrointestinal tumor in children is esophageal carcinoma. Suárez and colleagues reported the first case of a 14-year-old male adolescent with a history of intellectual disability and Barrett's esophagus who was admitted in February 2015 to the Instituto Nacional de Cancerología de Colombia and developed esophageal adenocarcinoma. Similar to our two patients with gastric adenocarcinoma, this adolescent presented with nonspecific gastrointestinal symptoms, such as dysphagia, weight loss, and emesis, leading to delayed diagnosis and consequently

complicating both treatment and prognosis. Unlike our cases, esophageal adenocarcinoma in adolescents appears to have a stronger association with environmental and inflammatory factors rather than genetic predisposition, and its therapeutic approach is also generally based on protocols designed for adults[17,18].

Furthermore, it is important to highlight that molecular testing was performed in our patients, allowing for better stratification and consideration of more specific treatments despite the current limitations of targeted therapies. However, rapid disease progression was observed, underscoring the need to consider diagnoses such as Lynch syndrome and HDGC when variants in the MSH2 and CDH1 genes are detected in young patients. This becomes especially relevant in the context of chemotherapy resistance, given the characteristic aggressiveness of these tumors in the pediatric population, even in the absence of a family history of cancer.

Consequently, targeted studies for pathogenic variants are recommended in first- and second-degree relatives, along with genetic and oncologic counseling[2,3]. In addition, HER2 and PD-L1 negativity currently limits access to targeted therapies, reinforcing the importance of performing early molecular studies and implementing personalized therapeutic strategies[16]. These strategies should include genetic screening in patients with a history of hereditary cancer and the development of new therapeutic alternatives adapted to the molecular characteristics of tumors in this age group[9].

Finally, the presence of *H. pylori* in one of the cases supports its possible role as a cofactor in predisposed individuals and in high-prevalence populations, suggesting that early eradication could represent a relevant preventive measure.

CONCLUSIONS

The management of gastric adenocarcinoma in pediatric patients remains limited and poorly understood because of the lack of specific guidelines and the reliance on adult treatment protocols. Advancing research, including genetic studies in patients and their families, is essential to improve knowledge and optimize treatment strategies in this population.

By presenting these cases, we hope to contribute to the literature regarding the importance of early diagnosis of gastric cancer in the pediatric population, based on the need to perform early diagnostic studies even in the presence of nonspecific symptoms such as hematemesis, abdominal pain, weight loss, or lumbar pain. These measures will facilitate early detection and the development of appropriate therapeutic strategies and, ultimately, improve the clinical prognosis of these patients.

Conflict of Interest

No conflicts of interest were reported.

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