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CASE REPORT | SMALL BOWEL

Extraosseous Ewing Sarcoma With Upper Gastrointestinal Bleeding

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ABSTRACT

Ewing sarcoma/primitive neuroectodermal tumor (ES/PNET) is a type of malignancy that usually appears as a tumor in the bone. However, in a few patients with ES/PNET, it can occur outside of the bone. Although extraosseous ES/PNET can appear in various parts of the body, involvement of small bowel is rare. If it does, it can present with vague abdominal pain and gastrointestinal bleeding. This case report presents a 28-year-old gentleman with extraosseous ES/PNET in the duodenum who experienced gastrointestinal bleeding as a presenting symptom.

KEYWORDS: Ewing sarcoma; primitive neuroectodermal tumor; extraosseous Ewing sarcoma; small bowel Ewing sarcoma

INTRODUCTION

Ewing sarcoma/primitive neuroectodermal tumor (ES/PNET) was first described by Stout in 1918 as a soft tissue malignancy arising from neuroectoderm.[1] These tumors are typically found in the metaphysis and diaphysis of long bones, ribs, and pelvis. ES/PNETs can occur outside the bones in around 12% of patients.[2] These extraosseous lesions usually arise from the chest (44%), retroperitoneum and pelvis (26%), extremities (20%), head and neck (6%), kidneys, esophagus, ovaries, prostate, and are rare in the small bowel.[3]

The case presented here is of an extraosseous Ewing sarcoma involving the duodenum and presenting with gastrointestinal bleeding.

CASE REPORT

A 28-year-old man with a history of chronic alcohol consumption and smoking with no comorbidities from Western Nepal presented to the emergency department with pain in the abdomen and occasional black tarry stool for 2 years with an increase in symptoms over the last 7 days. On examination, the patient had pallor along with a palpable, nontender globular mass of approximately 2 cm × 2 cm in size in the epigastrium.

The laboratory workup of the patient revealed severe iron deficiency anemia with hemoglobin of 7.9 g/dL. The esophagogastroduodenoscopy done in other center was suspicious for duodenal malignancy (Figure 1). Contrast-enhanced computed tomography (CT) scan of the abdomen and pelvis reported a large ill-defined heterogeneously enhancing mass (57.9 mm × 53 mm × 61.2 mm) in the lateral wall of the first part of duodenum extending to pancreaticoduodenal groove with surrounding soft tissue stranding. The mass protruded into the lumen of duodenum with eccentric narrowing, abutting the pancreas and branch of a superior mesenteric artery along with portal vein. There was loss of fat plane and possible infiltration into the head of pancreas (however, the exact origin of mass could not be determined) along with periportal, perigastric, and mesenteric lymphadenopathy (Figure 2). Repeat esophagogastroduodenoscopy done 2 weeks later revealed a bulge over the antrum with a non-negotiable stricture, and an endoscopic biopsy was not feasible.

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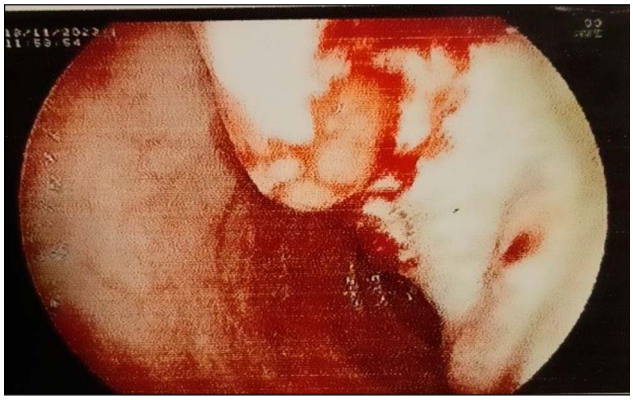

Figure 1. Esophagogastroduodenoscopy shows an ulcerating mass in the first part of the duodenum.

Endoscopic ultrasound showed a predominantly hypoechoic lesion measuring 55 mm × 65 mm compressing the antrum (Figure 3). A ultrasonography-guided trucut biopsy revealed small round cell tumor with a hyperchromatic nucleus, scattered mitosis, and necrosis (Figure 4). In immunohistochemistry study, tumor stained nuclear positivity for NKX2.2 and synaptophysin (rare cells) and negative for DESMIN, WT1, MYO D1, GATA3, CD45, and CK. Positron emission tomography-CT scan of whole body with contrast showed a large 18-flurodeoxyglucose (FDG) avid mass measuring 11.5 × 7 × 6.6 cm; maximum standardized uptake value (SUVmax) 27.5 involving the posterior wall of stomach, left lateral wall of the duodenum, and the head of pancreas as primary lesion; and multiple FDG avid perigastric, peripancreatic, periportal, portocaval (26 × 24 mm, SUVmax 11.8) lymph nodes and multiple FDG avid hypodense lesions, largest 30 × 28 mm, SUVmax 11 in segment VIII of liver suggestive of metastasis (Figure 5).

The case was referred to the medical oncology department for further management due to the nonfeasibility of en bloc resection. The patient was treated with a chemotherapy regimen

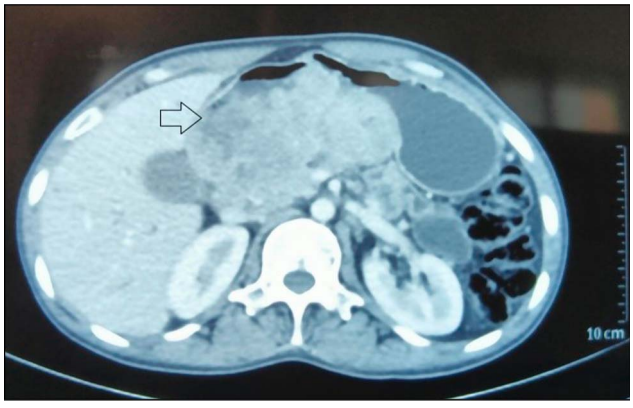

Figure 2. Computed tomography scan of the abdomen. Axial transverse view of large heterogeneously enhancing mass (57.9 mm × 53 mm × 61.2 mm) in the lateral wall of the first part of the duodenum (arrow).

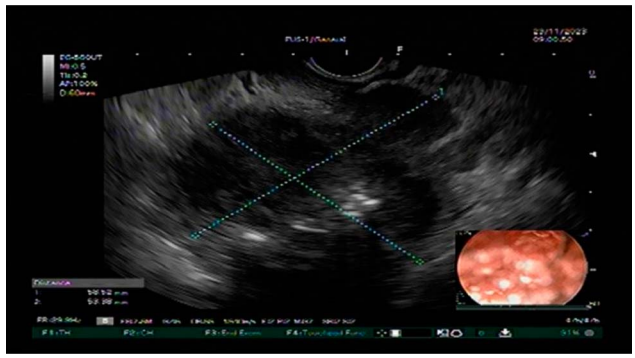

Figure 3. Endoscopic ultrasound shows a hypoechoic lesion measuring 55 mm × 65 mm compressing antrum.

of vincristine, doxorubicin, and cyclophosphamide along with hemostatic radiotherapy; however, the patient eventually succumbed to illness after 4 weeks of treatment.

DISCUSSION

ES/PNET usually appears as a tumor in the bone. However, in a few patients, it can present as an extraosseous tumor. Although extraosseous ES (EES) can appear in various parts of the body, involvement of the small bowel is rare. Case studies of extraosseous tumors involving the small bowel have reported the most common site as ileum (56%), followed by jejunum (33%) and duodenum (11%).[3–5] ES/PNETs show a male-to-female ratio of 1.4:1. Most cases occur in White individuals between the first and third decades of life, with the highest incidence in the second decade. However, patients with EES are 5–10 years older and have less predilections for men or Whites than patients with Ewing sarcoma of bone (ESB).[6] Histopathologically, ES/PNETs are uniformly distributed on small round cells with scant clear, eosinophilic cytoplasm, round nucleoli, and finely stippled chromatin.[7]

The 2 most common chromosomal translocations specific to ES/PNET are t(11;22)(q24;q12) and t(21;22)(q22;q12). Translocation of t(11;22)(q24;q12) is present in 90% of all cases and

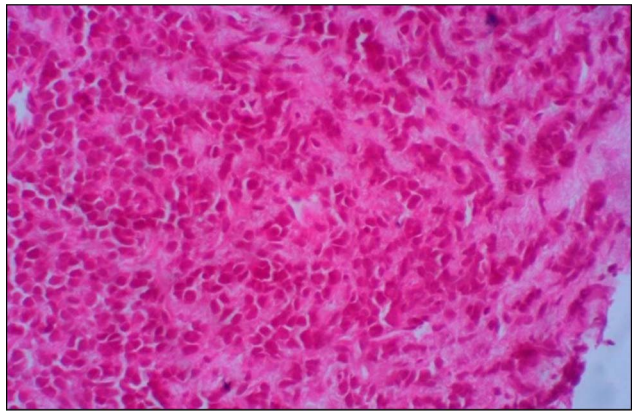

Figure 4. Histopathological examination shows sheets of small cells with round or oval nuclei and prominent nucleoli.

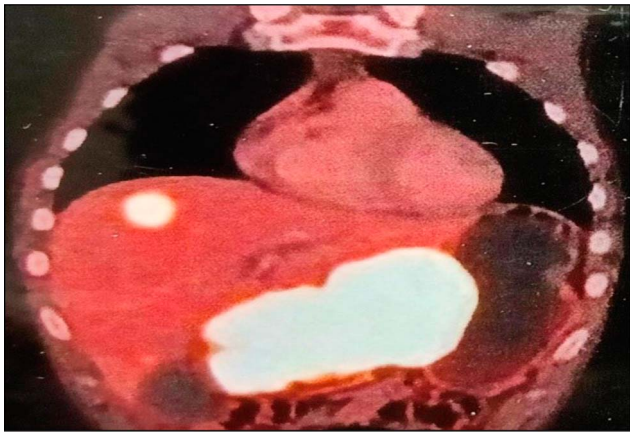

Figure 5. Positron emission tomography-computed tomography showing flurodeoxyglucose avid mass measuring 11.5 × 7 × 6.6 cm involving posterior wall of the stomach, left lateral wall of the duodenum, and head of the pancreas (primary lesion) and multiple flurodeoxyglucose avid hypodense lesions, largest 30 × 28 mm in segment VIII of the liver (metastatic lesion).

results in a fusion between the FLI1 gene on 11q24 and the EWSR1 gene on 22q12, creating an EWS/FLI-1 transcript. The resulting fusion product EWSR1-FLI1 functions as an oncoprotein and plays an important role in tumorigenesis.[8] CD99 positivity is observed in around 95% of cases, making it more sensitive but less specific.[9] By contrast, NKX2.2 positivity is more specific for ES/PNET.[10]

Duodenal ES/PNET is rare and may present with various symptoms, with the most common being nonspecific abdominal pain. Gastrointestinal bleeding may occur in approximately 15% of cases.[11] However, it may also present with intussusception, perforation, intestinal obstruction, and rupture.[4,11–13] Regardless of its origin, ES generally has a poor prognosis. Poor outcome determinants in duodenal ES include tumor size of more than 5 cm, old age, no response to chemotherapy, and the presence of metastasis.[14] Magnetic resonance imaging is preferred over CT for local staging of ES/PNET due to its high detection sensitivity for soft tissue contrast. However, FDG-positron emission tomography along with CT of the chest is used for detection of metastasis, as well as to assess chemotherapeutic response and detect recurrent disease.[15] A definitive diagnosis is made with a CT or US-guided core-needle biopsy or pathological examination of the resected surgical specimen.[16]

The National Comprehensive Cancer Network recommends local treatment (surgery and/or radiotherapy) along with chemotherapy for ES/PNET.[17] The current chemotherapy regimens involve alternating cycles of vincristine, doxorubicin, cyclophosphamide, and ifosfamide-etoposide every 2–3 weeks.[18] Radiotherapy is recommended only for inoperable lesions. The 5-year overall survival for localized EES is higher than for Ewing sarcoma of bone. The 5-year survival rate for localized ES/PNET is 65%–75% but for metastatic patients, it is <30%.[19]

To date, only 36 cases of small bowel Ewing sarcoma (including 4 cases of duodenal origin) have been reported. Given the rarity of this condition, we are reporting this case to contribute to the existing sparse literature.[4,5,12,20]

DISCLOSURES

Author contributions: AK Yadav, M. Shrestha, S. Rajbhandari, R. Kafle, S. Shrestha: acquisition, analysis and interpretation of data; drafting and reviewing of manuscript. D. Khadka, M. Poudel, and S. Poudel: analysis and interpretation of data; drafting and reviewing of manuscript. AK Yadav is the article guarantor.

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