



Primary duodenal embryonal rhabdomyosarcoma in adults: a case report

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Abstract: Rhabdomyosarcoma (RMS) is a soft tissue sarcoma that histologically resembles embryonic skeletal muscle. It is a rare malignancy in adults with a predilection in the head and neck, the genitourinary tract, and other extremities. However, RMS of the gastrointestinal tract is an even rarer condition that merits presentation and discussion. Here, we report on a case of primary duodenal embryonal RMS in an adult.

Keywords: Rhabdomyosarcoma (RMS); duodenum; sarcoma

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Introduction

Rhabdomyosarcoma (RMS) is a soft tissue sarcoma that histologically resembles embryonic skeletal muscle. It can occur anywhere in the body, including tissues devoid of skeletal muscles. The cell of origin of RMS remains unknown, however, recent evidence suggests that RMS can originate from aberrant development of non-myogenic cells (1).

Rhabdomyosarcoma is a common malignancy in children; indeed, it accounts for more than 50% of all soft tissue sarcomas in children (2). However, it is a rare malignancy in adults, where soft tissue sarcomas constitute less than 1% of all malignancies, and RMS accounts for only 3% of all soft tissue sarcomas (3). The most common locations in which RMS can be found are the head and the neck (35%), the genitourinary tract (22%), and other extremities (18%) (4). Most cases of gastrointestinal RMS represent a metastatic disease, with primary involvement being extremely rare. Primary RMS has primarily been reported from the oesophagus (5-8) and the stomach (9,10). Here, we report on a case of primary duodenal embryonal RMS in an adult.

Case presentation

A 79-year-old male with a known case of diabetes mellitus,

hypertension, and ischemic heart disease from 5 years ago, presented to the emergency room complaining of epigastric pain, nausea, vomiting, and weight loss. The vomiting had become intractable in the week before his presentation. He denied dysphagia, hematemesis, melena, hematochezia, diarrhea, and constipation.

Upon examination, he appeared chronically ill; however, his vital signs were normal. An abdominal exam revealed a soft abdomen with mild fullness and tenderness in the epigastric area, but without palpable masses. Other systems exams were unremarkable. Blood tests revealed only iron deficiency anaemia (haemoglobin 10 g/dL). Tumour markers, including carcinoembryonic antigen (CEA), CA19-9 and alpha-fetoprotein (AFP), were within the normal limits. A computed tomography (CT) scan (*Figure 1*) revealed a 7 cm × 9 cm mass arising from the duodenal bulb and invading the gastric antrum and pancreatic head. The mass caused high-grade gastric outlet obstruction. Moreover, multiple low-density lymph nodes (LN) were observed at the porta hepatis, peripancreatic, and retroperitoneal regions. The largest LN measured 24 mm at the porta hepatis. The liver showed multiple bilobar variable-sized hypodense lesions, which is suggestive of metastasis. Also, there was moderate free fluid in the abdomen and possible peritoneal metastasis.

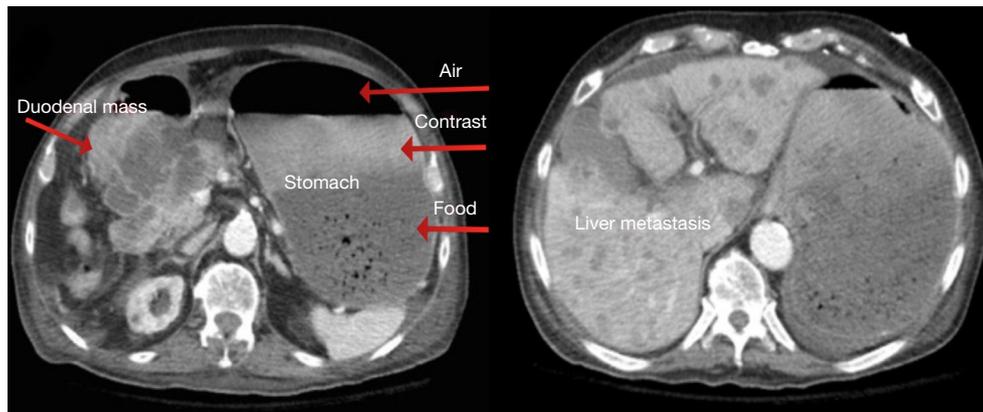


Figure 1 Duodenal mass invading pancreatic head and causing gastric outlet obstruction with liver metastasis.

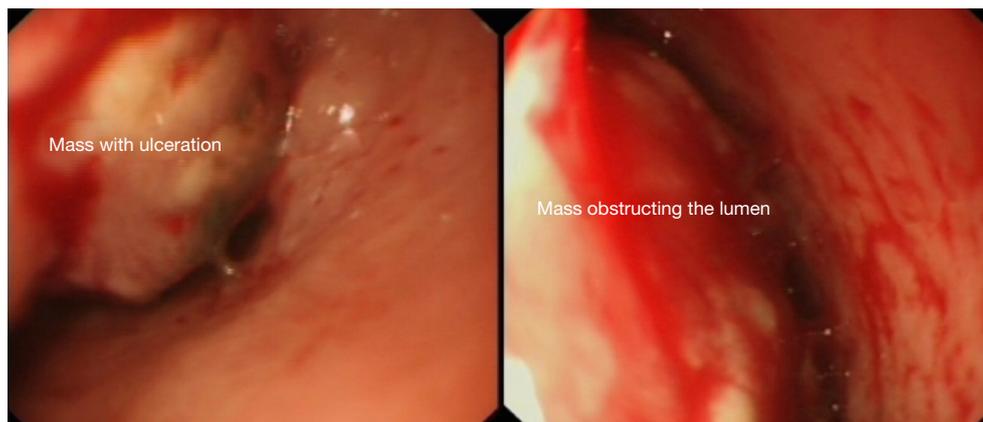


Figure 2 ulcerated mass originating from the duodenum and growing through the pylorus into the stomach causing gastric outlet obstruction.

An esophagogastroduodenoscopy (EGD) revealed a large irregular ulcerated mass originating from the duodenum and growing through the pylorus into the stomach, causing gastric outlet obstruction and preventing further examination of the duodenum (*Figure 2*). Multiple biopsies were taken for histopathology. A histologic examination revealed diffuse infiltration by a sheet of round, small blue cells (*Figure 3*). Moreover, areas with spindle-shaped and undifferentiated cells admixed with scattered round, strap, or tadpole-shaped eosinophilic rhabdomyoblasts were also present (*Figure 4*). Cytoplasmic cross-striation was identified as well. An immunohistochemistry and a molecular study revealed strongly and diffusely positive Desmin (*Figure 5A*) and Myogenin (*Figure 5B*). Moreover, smooth muscle actin (SMA) was focally positive (*Figure 5C*). A molecular study for FOXO1 (13q14) was conducted

by fluorescence in situ hybridization (FISH), but no rearrangement of the FOXO1 gene region was identified, precluding the possibility of alveolar-type RMS. Therefore, the final diagnosis was advanced duodenal embryonal RMS. The patient experienced poor functional status and planned for supportive care only. He passed away two weeks after admission, likely due to a massive pulmonary embolism.

Discussion

Primary malignant tumours of the duodenum represent 0.3% of all gastro-intestinal tract tumours, but can represent up to 50% of small bowel malignancies. The most frequent primary malignant tumour of the duodenum is adenocarcinoma (11). Other primary tumours include lymphomas, leiomyosarcomas, carcinoid tumours,

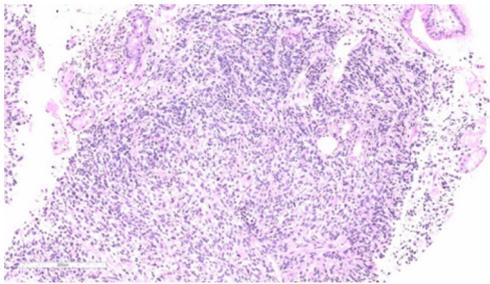


Figure 3 The infiltrate composed of small and round cells with mixed spindle shaped and strap shaped rhabdomyoblast (H&E, 10×).

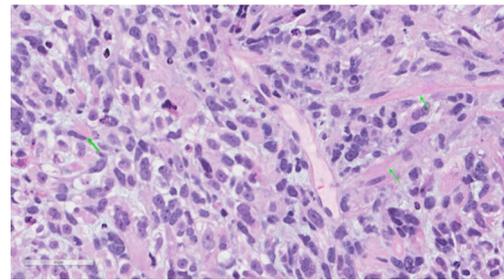


Figure 4 Spindle shaped and strap rhabdomyoblast with observed striation (arrows) seen on higher magnification (H&E, 40×)

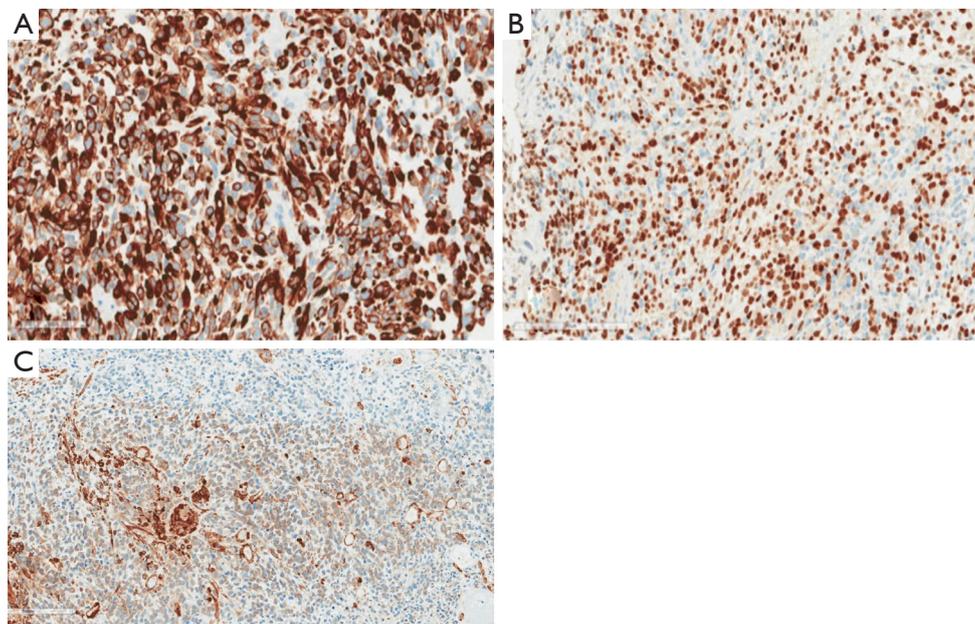


Figure 5 Strong and diffuse positive immunohistochemical stains for desmin (A) and myogenin (B). Focal positivity was noted in smooth muscle actin (SMA) (C). All figures magnification is 40×.

gastrinomas, and stromal tumours. These tumours usually present with bleeding or gastric outlet obstruction.

Duodenal RMS is an extremely rare malignancy. Through an extensive search of the relevant literature, we found only four case reports written in English that describe primary duodenal RMS in adults (*Table 1*). Rhabdomyosarcoma has been classified by the WHO into four histologic subtypes: embryonal, alveolar, pleomorphic, and spindle cell/sclerosing RMS (16). In adults, the most common histologic subtype of RMS is the pleomorphic subtype (17). Our case is the first reported case of embryonal subtype duodenal RMS in an adult.

Although RMS is considered a single disease, there are important differences in how these tumours behave depending on tumour location, histologic subtype, tumour bulk, patient’s age at the time of diagnosis, and the presence or absence of distant metastasis. Because there are so few reported cases of duodenal RMS, it is not possible to speculate on its behaviour, response to treatment, or prognosis. However, RMS outcomes are considerably less favourable for adults than for children (18). It seems that the pleomorphic subtype carries the worst prognosis (17). Distant metastasis at the time of diagnosis is quite common and the lung is the most common site for metastasis (18).

Table 1 Reported cases of duodenal rhabdomyosarcoma (RMS)

No	Reference	Age (y)	Sex	Presentation	Histology	Metastasis	Treatment	Prognosis
1	Moses <i>et al.</i> (12)	66	F	Persistent Fever	NA	Liver	NA	NA
2	Kazuki <i>et al.</i> (13)	68	M	abdominal pain, anorexia, weight loss	NA	None	Surgery	NA
3	Sato <i>et al.</i> (14)	63	M	Melena	Pleomorphic	None	Surgery	NA
4	Shimamura <i>et al.</i> (15)	73	F	Severe anemia	Pleomorphic	None	Surgery	No recurrence within 6 months
5	Alkhormi <i>et al.</i>	79	M	Abdominal pain, nausea, vomiting, weight loss	Embryonic	Liver and lung	Conservative	Died

Please note that the 5th case is the current case report. NA, not available.

Invasion of the pancreatic head and metastasis to the liver are probably common in RMS arising from the duodenum.

The optimal management of RMS in adults is unknown due to its rarity and the absence of any standard treatment protocol or guidelines. The current treatment standards for adult RMS follow the treatment standards created for children, which were proposed by the Intergroup Rhabdomyosarcoma Studies (IRS) (19). These treatment standards include multimodal therapy (MMT: resection, chemotherapy, and radiation). Surgery is the mainstay of treatment for adult RMS, as it has been correlated with an improved survival rate (20). According to the IRS protocols, all RMS patients should undergo radiotherapy to achieve long-term local control of the tumour (21). The IRS protocols also recommend that all RMS patients receive combination chemotherapy as soon as a diagnosis is made, or after resection, because this significantly improves survival rates. The recommended regimen is a combination chemotherapy approach consisting of vincristine, actinomycin, etoposide or ifosfamide, and cyclophosphamide. The three-year, failure-free survival rate ranges from 78–92% with these regimens (22). The treatment of duodenal RMS should follow the treatment standards of RMS elsewhere in the body until more cases have been evaluated to uncover further clinical characteristics, and thus determine the optimal management for this tumour.

Acknowledgments

None.

Footnote

Conflicts of Interest: The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Written informed consent was obtained from the patient for publication of this manuscript and any accompanying images.

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