Perivascular Epithelioid Cell Neoplasm (PEComa) as a Cause of Abdominal Pain in a Child

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Perivascular epithelioid cell neoplasm (PEComa) is a very rare mesenchymal tumor associated with perivascular epithelioid cell formation that commonly presents in a perivascular location. PEComas may present at the falciform ligament, the hepatic right lobe, as well as other locations. They are generally associated with minimal symptoms although abdominal pain may be present as an initial symptom. We present a case of a PEComa of the falciform ligament leading to abdominal pain in a young male.

A 10 year-old previously healthy male presented with a chief complaint of epigastric pain and gastroesophageal reflux symptoms for two years. The abdominal pain was intermittent in nature. Although he complained of intermittent gastroesophageal reflux, he denied emesis, hematemesis, bilious emesis, dysphagia, or odynophagia. He had no weight loss or early satiety. In the gastroenterology clinic, the patient was noted to have normal vital signs and growth parameters (per CDC growth charts). His physical exam was normal. Laboratory testing revealed a normal complete blood count and hepatic function panel. An erythrocyte sedimentation rate, alpha fetoprotein level, lactate dehydrogenase level, and uric acid level were normal.

An esophagogastroduodenoscopy (EGD) was performed which was normal except for evidence of a raised area consistent with a possible mass near the antrum with no overlying mucosal changes (Figure 1). Forcep biopsies of this area were normal. A subsequent computed tomography scan of the abdomen revealed a round soft tissue mass (measuring 1.8 x 2.4 x 2.2 centimeters) in an anterior aspect of the fissure of the falciform ligament (Figure 2). This lesion was exerting a mass effect on the lesser curvature of the stomach. An endoscopic ultrasound (EUS) was performed which revealed a round soft tissue mass (measuring 1.8 x 2.4 x 2.2 centimeters) in an anterior aspect of the fissure of the falciform ligament (Figure 2). This lesion was exerting a mass effect on the lesser curvature of the stomach. An endoscopic ultrasound (EUS) was performed which revealed a 2.4 centimeter, heterogeneous lesion that was round to oval in shape and well demarcated. The lesion was densely hypervascular, with extensive Doppler flow activity along the entire periphery of the lesion and, to

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a lesser extent, within the lesion itself. The lesion did not appear to arise from the gastric wall, and there was no peri-lesional adenopathy (Figure 3).

The patient underwent laparoscopic surgical excision, and a spherical mass was found within the falciform ligament without evidence of peritoneal implants. The liver and gallbladder appeared normal. The mass was shown to measure 2.8 x 2.4 x 1.7 centimeters (weight 6.9 grams), and it consisted of an encapsulated purple-tan soft tissue mass with attached fibromembranous tissue (Figure 4). On microscopic examination, the mass demonstrated a fascicular and nested proliferation of spindle to epithelioid cells with clear to eosinophilic cytoplasm and round to oval nuclei (Figure 5). There was no mitotic activity, necrosis, or vascular invasion identified. Immunohistochemically, the cells were positive for smooth muscle marker (smooth muscle actin) and melanocytic markers (HMB-45 and Melan-A) (Figures 6 and 7). Cell staining was negative for AE1/AE3, EMA, desmin, MYF-4, S-100, ALK-1, CD117, and CD34. These findings were consistent with a PEComa of the falciform ligament.

The pediatric oncology service was consulted on this patient, and it was decided that the risk of tumor recurrence and metastases was low. He has been scheduled for annual abdominal ultrasounds. Interestingly, all symptoms of abdominal pain and gastroesophageal reflux resolved after tumor removal.

DISCUSSION

PEComas neoplasms are characterized by perivascular epithelioid cells with a myomelanocytic immunophenotype that are typically arranged around the vasculature.\(^1,2,3\) These cells have a characteristic epithelioid or spindled appearance, and such cells that express both smooth muscle and melanocytic cell markers using immunohistochemistry staining.\(^4\) Many PEComas are benign, but they also have been reported to be malignant. The ratio of PEComa formation is equivalent between males and females in prepubertal children, although there is a strong female predominance of PEComa formation in adolescents and adults.\(^5\) There is a known association of PEComa with tuberous sclerosis.\(^6,7\)

The differential diagnosis of submucosal gastric masses is quite large but will include lipomas, duplication cysts, pancreatic rests, PEComas, gastrointestinal stromal tumors, metastatic melanoma, clear cell sarcoma, leiomyosarcoma, carcinoid tumor as seen in multiple endocrine neoplasia type 1, and paragangliomas.\(^8\) These lesions may be difficult to differentiate based on morphology alone although use of endoscopic ultrasound and tissue immunohistochemistry greatly aids in the diagnosis.\(^5,9,10\)

Notably, clear cell sarcomas of a gastrointestinal variant

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consist of spindle and epithelioid cells with diffuse S100 positivity and variable positivity for other melanocytic markers and can be confused with a PEComa based on immunohistochemistry. Molecular genetic studies to demonstrate the EWS-ATF fusion gene representing t(12;22) q13;12) translocation or the EWS-CREB3L2 fusion gene representing t(2;22) q34;12) translocation can be used to differentiate clear cell sarcoma from PEComa a clear cell sarcoma is often S100 positive and may be difficult to differentiate from PEComa based on immunohistochemistry.\(^\text{10,11,12}\)

Previously, tumors such as angiomyolipoma, lymphangioleiomyomatosis, primary extrapulmonary sugar tumor, and clear cell myomelanocytic tumor were noted to have similar histologic characteristics although they were present in different anatomic locations. Hence, the term “PEComa” has been developed to broadly characterize this tumor grouping with specific histologic findings regardless of anatomic location or morphology.\(^\text{5,13}\) As a result, PEComas have been described as occurring at the kidney, liver, lung, nose, bladder, and other locations.\(^\text{1,13,14,15,16}\) PEComas have been noted to occur in the gastrointestinal tract anywhere from the stomach to the rectum. PEComas of the gastrointestinal tract may either tend to be located in the serosa with involvement of all associated bowel layers or will be polypoid tumors involving the mucosa and submucosa.\(^\text{8,17}\) In our described case, the presumed gastric mass was, in actuality, a gastric compression from a PEComa at the falciform ligament which is a known location for such a tumor.\(^\text{18}\)

PEComas are rare in children, and approximately 40 pediatric cases have been documented as occurring in the pelvis, vagina, eye/orbit, and gastrointestinal tract (including the duodenum, appendix, colon, and rectum).\(^\text{5}\) Abdominal pain may be a presenting symptom in a patient with an abdominal PEComa.\(^\text{1}\) In 2000, a unique pediatric case series of 7 patients described PEComas occurring exclusively at or near the ligamentum teres and falciform ligament. Most of these cases occurred in females, and the cases occurred at an average age of 11 years. Immunohistochemistry staining demonstrated that the tumors were positive for smooth muscle actin, melin-A, and myosin but were negative for desmin, in a pattern similar to our patient. Follow-up data, available in six of seven cases, showed five patients to be free of disease, and one to have a radiographically presumed lung metastases.\(^\text{19}\)

PEComas have been described as benign or malignant although the criteria for malignancy are not well defined. Most PEComas located at the falciform ligament or ligamentum teres are benign, and the therapy

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for such tumors is surgical resection.\(^5\) PEComa features associated with a higher risk of malignancy include large size (greater than 5 centimeters), infiltrative growth, mitoses greater than 1 in 50 per high power field, vascular invasion, and necrosis.\(^20\) The PEComa of our described patient had a diameter less than 5 centimeters and had no significant nuclear atypia, necrosis, or mitotic activity likely consistent with a benign lesion.

Surgical resection is the treatment of choice for most PEComas; however, chemotherapy may be indicated for PEComas with histologic features consistent with malignant disease or in the event of associated metastases.\(^5,21\) Doxorubicin and ifosfamide have been used as chemotherapy for PEComas, and rapamycin, an inhibitor of m-TOR, also has been used as treatment for malignant PEComa.\(^22,23\) Close follow-up with serial imaging (such as ultrasound) is necessary to evaluate for PEComa recurrence.\(^24\)

**References**


**Answers to this month’s crossword puzzle:**

![Crossword Puzzle Image]