Perivascular Epithelioid Cell Tumor Presenting as a Presacral Mass

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Background	The differential diagnosis for tumors originating in the retrorectal space is vast and ranges from benign congenital cysts to malignant neurogenic tumors. Perivascular epithelioid cell tumors (PEComas) are rare, mesenchymal neoplasms with distinct immunohistochemistry staining and can appear in various locations with kidney and liver being most common sites.
Summary	We present a 66-year-old female who was found to have an incidental presacral mass. This was treated with complete surgical resection. Histopathology revealed a clear cell neoplasm embedded in well-vascularized fibroconnective tissue with immunohistochemical staining positive for HMB-45 and PNL2 expression, diagnostic of PEComa.
Conclusion	The presacral space represents a histologically diverse location with a wide array of potential pathology. A standardized approach to the work-up and treatment of presacral masses adhering to sound oncologic principles will provide the best outcome.
Keywords	PECOMA, presacral, retrorectal, perivascular, epithelioid, mass

DISCLOSURE:
The authors of this paper have no conflicts of interest to disclose.

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Case Description

A 66-year-old female with a medical history of hypertension and asthma was incidentally found to have a presacral mass on cross-sectional imaging during a work up for abdominal pain. Rectal examination was significant for a smooth, round mass without overlying mucosal abnormalities, with the superior most extent of the lesion easily palpable with the examining digit. The physical examination was otherwise unremarkable. A computed tomography (CT) scan demonstrated a 4.4 cm presacral mass lesion (Figure 1). This was followed by a magnetic resonance imaging (MRI) scan for further characterization, which revealed a mixed cystic and solid presacral mass on T2-weighted imaging (Figure 2).

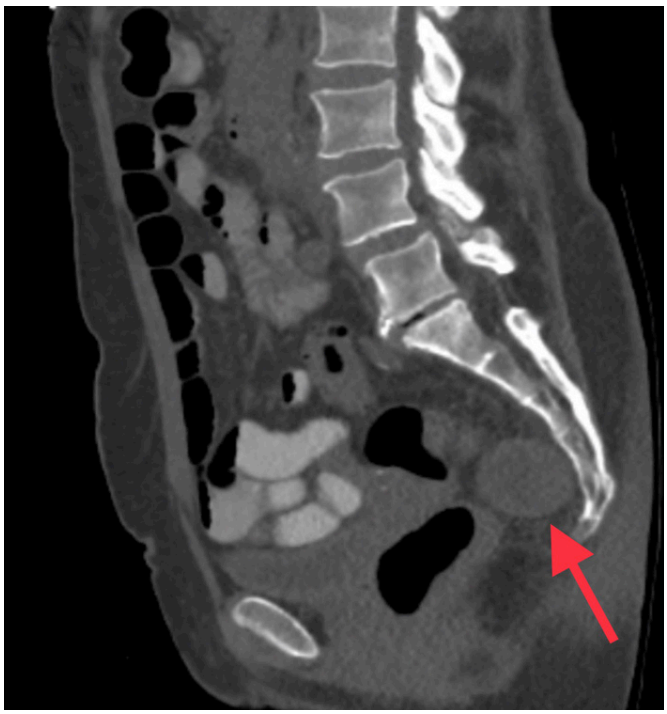

Figure 1. CT Saggital section demonstrating 4.4 x 3.6 cm presacral mass

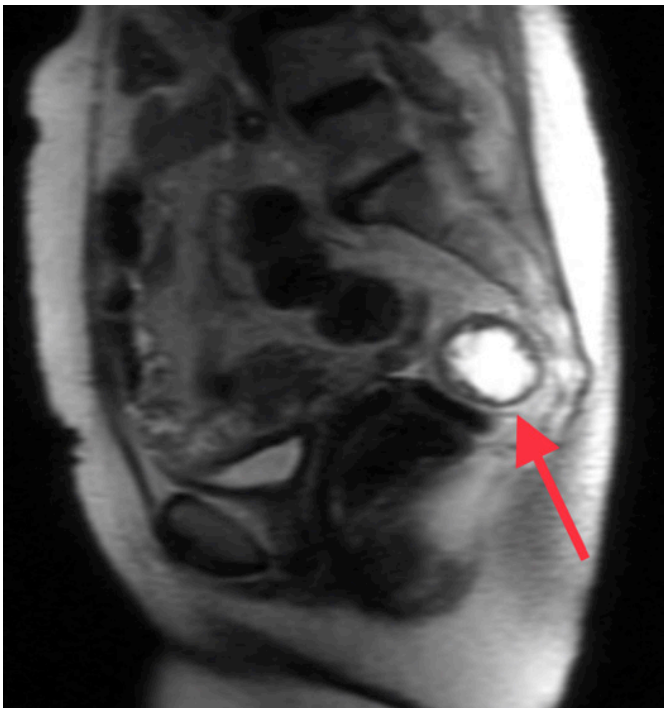

Figure 2. MRI T2 weighted image; sagittal section demonstrating 4.1 x 3.6 x 3.5 cm presacral cystic lesion with surrounding enhancing solid soft tissue. Lesion is separate from the rectum and surrounding bones

The patient underwent an elective excision of the mass via a posterior, trans-coccygeal (Kraske) approach. This was accomplished through a vertical para-sacral incision. Dissection was carried down through the subcutaneous tissue until the coccyx and anococcygeal ligament were visualized. Excision of the coccyx was necessary for adequate exposure. The operating assistant's digit was placed in the rectum throughout the entirety of the procedure for easy identification of the rectal wall and for the application of caudad traction to the mass. The lesion was removed in its entirety without injury to the rectum or surrounding structures. A drain was placed and the wound was closed in multiple layers.

Histopathologic examination revealed a clear cell neoplasm embedded in well-vascularized fibroconnective tissue. The stromal tissue contained a mixed inflammatory infiltrate with patchy hemorrhage. The tumor cell grew in a nested pattern, surrounded by fibrous septae (Figure 3). High-power evaluation demonstrated epithelioid cells with well-defined cell borders. The tumor cell cytoplasm was predominantly clear, with focal collections of amorphous eosinophilic material. Nuclei were of variable size and shape. Most contained prominent nucleoli. Mitotic figures were not well demonstrated (Figure 4). Supplemen-

tal immunohistochemical staining showed HMB 45 and PNL2 expression. These findings were consistent with the diagnosis of a perivascular epithelioid cell tumor (PEComa).

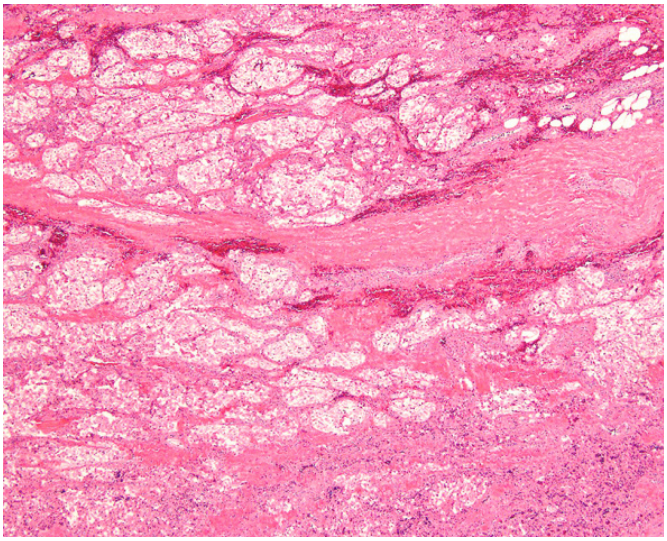

Figure 3. Low Power Magnification 10x

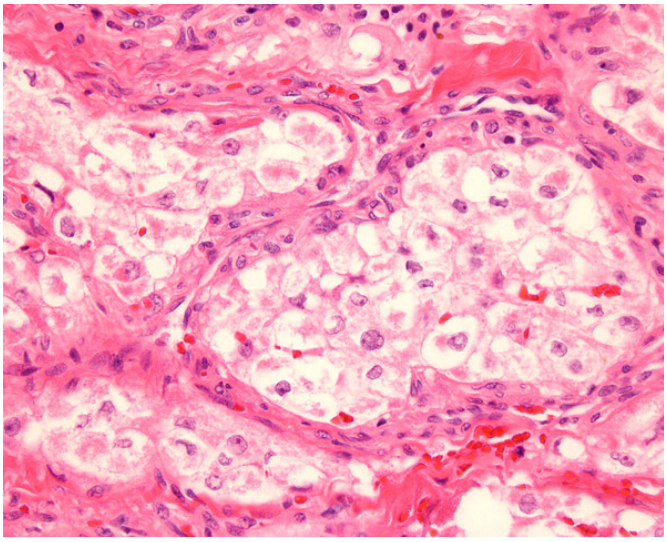

Figure 4. High-power magnification 40x.

Discussion

The retrorectal space is found between the presacral fascia and the fascia propria which envelopes the mesorectum. The differential diagnosis for tumors originating in this space is vast and ranges from benign congenital cysts to malignant neurogenic tumors.[1] Symptoms can be vague and the majority of masses are found incidentally on rectal examination.[2] Cross-sectional imaging is essential to determine the appropriate operative approach for excision. Preoperative biopsy is controversial and is generally omitted. Due to the possibility of malignancy, it is recommended that all presacral masses undergo excision.[1,3] The surgical approach itself (transabdominal, transcoccygeal, or a combination of both) depends on the location and extent of the mass. We present a case of a PEComa found in the presacral space, a unique location not previously described in the literature.

Perivascular epithelioid cell tumors (PEComas) have been described in previous case reports but little is still known about these tumors due to their extreme rarity. First described in 1992 by Bonetti, PEComas are characterized as mesenchymal neoplasms composed of distinct perivascular epithelioid cells that are immunoreactive for melanocyte and smooth muscle markers.[4] Due to extremely rare incidences, there are no unified guidelines on characteristic imaging, diagnostic work-up, clinical presentation, and management of PEComas. Previous case reports identified the tumor in various locations including kidney, liver, gynecologic organs, retroperitoneal soft tissues, lungs, palate, and left groin.[4,5] Consensus on most common locations for development of PEComa is lacking due to the wide variety of subtypes and lack of large-scale, unified studies. A review conducted by Tan et al identified the kidney as the most common location for PEComas in 32 cases (16 of 32).[4] Other common locations have been reported to be liver, colon, and uterus.[5,6] We found only one other mention of a presacral PEComa, which was reported by Messick et al.; however, no additional details on the PEComa were provided.[7] Risk factors for development of PEComas remain elusive. Association with tuberous sclerosis complex (TSC) has been reported by Prasad et al.[6] Recent advances in genetic studies support this association as non-TSC PEComas have also been found to have changes in the TSC genes. However, additional studies are required to elucidate the exact pathogenesis.[8]

Current mainstay treatment of PEComas is complete surgical resection with adequate margins of healthy tissue, which is considered curative.[4] If the PEComa mass shows signs of malignancy such as high mitotic activity, atypical mitotic figures, coagulative necrosis, pleomorphism and nuclear atypia, infiltrative growth patterns, large size, local recurrence or distant metastases, chemotherapy is considered.[5]

Conclusion

In conclusion, the presacral space represents a histologically diverse location with a wide array of potential pathology. A standardized approach to the work-up and treatment of presacral masses adhering to sound oncologic principles will provide the best outcome.

Lessons Learned

Tumors arising from the presacral space have a vast differential diagnosis including both benign and malignant neoplasms. Biopsy of these lesions is not required prior to resection. Adhering to strict oncologic principles from work up to resection is critical to minimize the risk of recurrence.

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