Colonic Merkel Cell Carcinoma of Unknown Primary: Small Cell Carcinoma Outside the Usual

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Case Presentation and Management

- A 69-year-old female with history of bilateral ductal carcinoma of the breast, pulmonary adenocarcinoma, and anal squamous cell cancer presented with abdominal pain, distention, and reduced bowel movements. Computed tomography demonstrated dilated loops of small bowel with a partial obstruction at the level of the hepatic flexure.
- Colonoscopy revealed a 4cm necrotic mass at the hepatic flexure and a strictured area through which the scope could not pass. Biopsy revealed a high-grade neuroendocrine tumor.
- She underwent a right hemicolectomy and surgical specimen confirmed small cell neuroendocrine carcinoma, involving the entire wall of the colon extending into the pericolic adipose tissue to the radial margin, with serosal and subserosal deposits of small cell neuroendocrine carcinoma.
- Ki-67 immunohistochemical stained tissue showed >98% proliferative index with many foci of tumor necrosis and mitotic figures. The malignant cells stained positive on keratin CAM 5.2 and synaptophysin and negative on CDX-2 and TTF-1, suggestive of a non-colonic, non-pulmonary primary lesion of neuroendocrine origin.
- 14 of 17 tested lymph nodes stained positive for metastatic small cell neuroendocrine carcinoma, many with extracapsular spread.
- Merkel cell polyomavirus immunohistochemistry positive.
- Dermatologic evaluation revealed no primary lesion of MCC.

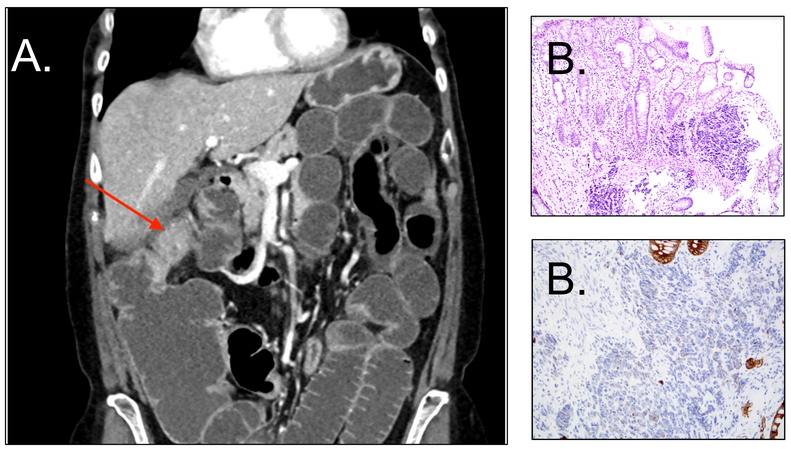

Figure 1. A. Computed tomography of the abdomen demonstrating dilated loops of small intestine suggestive of large bowel obstruction. Right colon mass visible at the red arrow. B. Histology showing malignant small round blues cells in the submucosa and mucosa, positive for keratin AE1/AE3, confirming carcinoma.

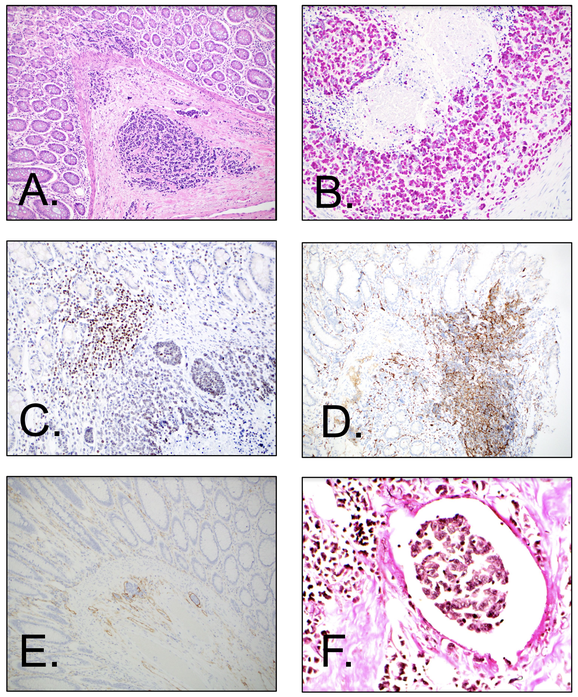

Figure 2. A. Submucosal and mucosal invasion of colon B. Ki-67 confirming high proliferative index. C. Merkel cell polyomavirus positive staining of malignant neuroendocrine carcinoma. D. Synaptophysin positive malignant cells. E. D2-40 demonstrating lymphatic invasion. F. Verhoeff's stain demonstrating venous invasion.

Discussion

Merkel cell carcinoma (MCC) is a rare neuroendocrine carcinoma of primary cutaneous origin which commonly metastasizes to the liver, lungs, and gastrointestinal (GI) tract.[1] Tumors that lack dermatologic primary lesions are dubbed Merkel Cell Carcinoma of Unknown Primary Origin (MCCUP).[2] Prior cases of MCC metastatic to the colon are rare, but all have been reported with known primary lesions elsewhere.[3] Confirmation of metastatic MCCUP to the colon is difficult due to its low incidence, lack of primary lesion, and varied clinical presentation.

It is important to differentiate MCCUP from other types of cancers that more commonly metastasize to the GI tract, including pulmonary or gastrointestinal small cell carcinoma, lymphoma, and metastatic undifferentiated anaplastic carcinoma. Merkel cell polyomavirus is present in over half of MCC tumors, especially in patients with an extensive cancer history or who are immunocompromised.[5]

Conclusions

Our case is the first describing MCCUP of the colon, highlighting the need for provider awareness of Merkel cell carcinoma as a root cause of neuroendocrine tumors of the gastrointestinal tract. In the setting of hepatic flexure mass, providers should consider Merkel cell carcinoma in their differential, even without skin findings, as a potentially insidious malignancy.

References

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