A Rare Case of Sporadic Metastatic Rectal Small Cell Carcinoma with Primary Biliary Cholangitis

Ans Albustamy MD[1], Arturo Rivera Suplee MD[2], Prateek Harne MD[2], Asif Zamir MD[2]
[1]Department of Internal Medicine, University of Texas Rio Grande Valley School of Medicine-Doctors Hospital at Renaissance; Edinburg, Texas
[2]Division of Gastroenterology, University of Texas Rio Grande Valley School of Medicine-Doctors Hospital at Renaissance; Edinburg, Texas

INTRODUCTION

Small cell carcinoma (SmCC) of the gastrointestinal tract (GIT) is a rare and highly aggressive malignancy, with an estimated prevalence of 0.1% to 1% of all gastrointestinal (GI) tumors[1]. At this time, the only identified risk factor is a positive family history[2]. Otherwise, no known association exists between SmCC and other conditions in the literature[3]. We present a patient with a history of primary biliary cholangitis and SmCC of the GIT, a rare cause of liver cirrhosis thought not to be associated with malignancy.

CASE DESCRIPTION

A 49-year-old woman with biopsy-proven PBC on ursodiol therapy was admitted due to three months of worsening rectal pain and bleeding. Liver chemistries revealed AST 146 IU/L, ALT 122 IU/L, ALP 1,106 IU/L, and total bilirubin 4.7 mg/dL. A CT scan showed multiple liver and pulmonary nodules concerning for metastatic disease in addition to an ill-defined mass in the rectum. PET scan confirmed these findings.

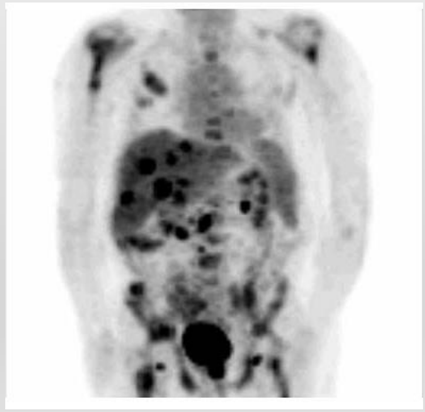

Image 1. PET scan revealing rectal mass, pleural involvement and liver lesions

CASE CONTINUED

MRCP confirmed the innumerable liver lesions with normal caliber biliary ducts. Colonoscopy showed a large, ulcerated rectal mass without obstruction but narrowing of the rectal vault, biopsy was strongly positive for synaptophysin, chromogranin, INSM, CD56, Villin and SATB2 suggestive of poorly differentiated neuroendocrine carcinoma, small cell type, with intact staining for MLH1, MSH2, MSH6 and PMS2 showing no evidence of mismatch repair.

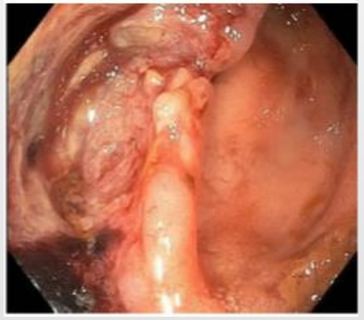

Image 2. rectal mass on initial colonoscopy

Patient was discharged in stable condition and started chemotherapy for metastatic SmCC with carboplatin/etoposide (Atezolizumab was not available as it was not approved by insurance). Unfortunately, our patient expired due to complications of chemotherapy.

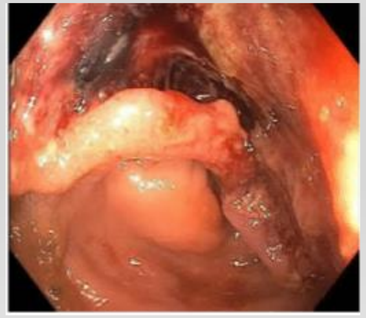

Image 3. ulcerated rectal mass on sigmoidoscopy

DISCUSSION

Our case reports a clinical and morphological presentation of a rare sporadic small cell carcinoma with mixed neuroendocrine features in a patient with PBC. Only a handful of cases exist in literature describing small cell neuroendocrine tumors, and all appear to have very poor prognosis, typically occurring in older patients[4]. Hindgut neuroendocrine tumors are typically asymptomatic until they obstruct or metastasize and are often found incidentally on colonoscopy. An age-appropriate colonoscopy in our case could have led to better outcomes, hence emphasizing its importance. It was interesting to note the history of PBC in our patient, but an association between PBC and SmCC has not been well studied. As opposed to primary sclerosing cholangitis that is well established to be linked with colorectal cancer, PBC has only recently been linked with an overall increased likelihood of all cancer[5], and more research is needed to establish a relation with SmCC.

REFERENCES

[1] Brenner, B., Tang, L. H., Shia, J., Klimstra, D. S., & Kelsen, D. P. (2007, February). Small cell carcinomas of the gastrointestinal tract: clinicopathological features and treatment approach. In *Seminars in oncology* (Vol. 34, No. 1, pp. 43-50). WB Saunders.

[2] Brenner, B., Shah, M. A., Gonen, M., Klimstra, D. S., Shia, J., & Kelsen, D. P. (2004). Small-cell carcinoma of the gastrointestinal tract: a retrospective study of 64 cases. *British journal of cancer*, *90*(9), 1720-1726.

[3] Brenner, B., Tang, L. H., Klimstra, D. S., & Kelsen, D. P. (2004). Small-cell carcinomas of the gastrointestinal tract: a review. *Journal of Clinical Oncology*, *22*(13), 2730-2739.

[4] Galanis, E., Frytak, S., & Lloyd, R. V. (1997). Extrapulmonary small cell carcinoma. *Cancer: Interdisciplinary International Journal of the American Cancer Society*, *79*(9), 1729-1736.

[5] Liang, Y., Yang, Z., & Zhong, R. (2012). Primary biliary cirrhosis and cancer risk: A systematic review and meta-analysis. *Hepatology*, *56*(4), 1409-1417.