Malignant Extrarenal Rhabdoid Tumor (MERT) of the Colon

by Deepti Dhavaleshwar, Kofi Clarke

Ten cases of primary colonic malignant extrarenal rhabdoid tumors (MERT) have been reported. We report a case in a 31-year-old female who presented with abdominal pain, nausea and vomiting. Her initial evaluation was significant for leukocytosis and an elevated erythrocyte sedimentation rate. On colonoscopy, a 4-cm localized inflammatory-appearing lesion at the ileocecal valve was noted. Biopsies showed poorly differentiated adenocarcinoma. Surgical pathology after right hemi-colectomy revealed a malignant neoplasm with rhabdoid features. Despite aggressive chemotherapy, the patient died four months after diagnosis. MERT cases are rare and have poor prognosis. A better understanding of these tumors may help to improve outcomes.

BACKGROUND

Malignant extrarenal rhabdoid tumors (MERT) of the gastrointestinal tract are extremely rare. To our knowledge, only 10 cases of primary colonic MERT have been reported.[1-9] We report a case of MERT involving the colon in a 31 year-old female patient.

CLINICAL CASE

A 31-year-old Caucasian female with no significant past medical or surgical history presented with right lower quadrant abdominal pain, nausea and vomiting. The pain was non-radiating, rated 10/10 in intensity and had no aggravating or relieving factors. She denied diarrhea, constipation, loss of appetite or weight changes. Her family history was significant for Crohn's disease in a maternal aunt.

Deepti Dhavaleshwar, MD; Kofi Clarke, MD, FRCP, Allegheny Health Network, Allegheny General Hospital, Division of Gastroenterology, Pittsburgh, PA

Initial work up was significant for leukocytosis of 14,000 cells/mL (normal range: 4,000-10,000 cells/mL) and an elevated erythrocyte sedimentation rate of 44 mm/hr (normal range: 0-20 mm/hr). Computed tomography (CT) scan of the abdomen and pelvis with contrast showed small bowel obstruction (SBO), focal thickening of the terminal ileum, mesenteric engorgement and upstream inflammation with skip lesions in the distal ileum (Figure 1). Colonoscopy performed after resolution of the SBO showed a 4-cm localized inflammatory-appearing lesion at the ileocecal valve. The terminal ileum could not be intubated because of partial obstruction (Figure 2). Biopsies showed poorly differentiated adenocarcinoma (Figure 3), and the patient underwent a right hemi-colectomy. Surgical pathology revealed a poorly differentiated malignant neoplasm with rhabdoid features, and 9 out of 15 pericolonic lymph nodes were positive for metastatic malignancy. Immunostains showed the tumor cells were positive for cytoketain AE1/AE3, negative

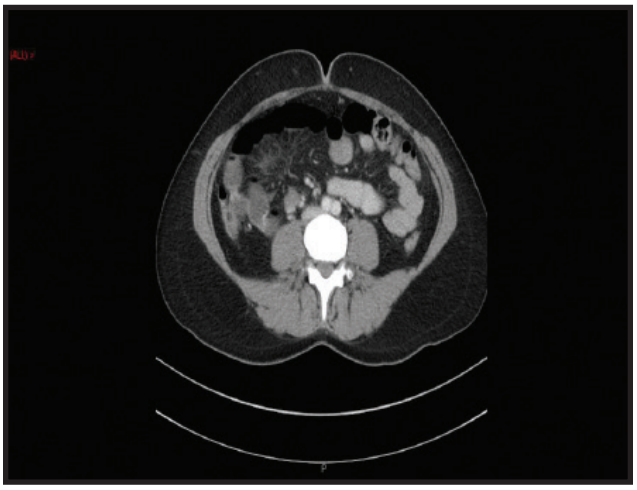

Figure 1. CT scan showing terminal ileum thickening.

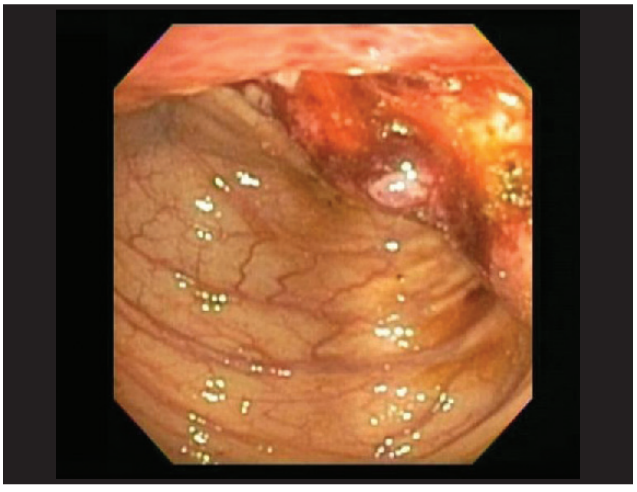

Figure 2. Cecal mass seen on colonoscopy.

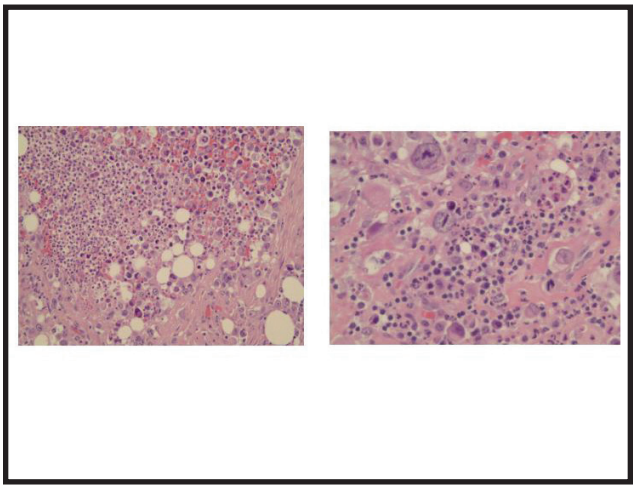

Figure 3. Pathology of surgical specimen.

for CK7, CK20 and CDX-2. Post-operative workup including positron emission tomography (PET) CT revealed multiple fluorodeoxyglucose (FDG) avid hepatic lesions, consistent with metastatic disease. The patient underwent chemotherapy with FOLFOX (a combination of leucovorin calcium [folinic acid], fluorouracil, and oxaliplatin), but despite the aggressive therapy, the patient succumbed to the disease four months after diagnosis.

DISCUSSION

The term malignant rhabdoid tumor (MRT) was originally used to describe a variant of pediatric renal tumor, which was clinicopathologically distinct from Wilms tumors. MRT of the kidney demonstrates a particularly aggressive growth and poor prognosis.[10] Malignant tumors with similarly appearing rhabdoid cells were subsequently described at extrarenal sites, and these have been described as MERT.

There appears to be no sex predilection and overall survival is less than 12 months. MERT is derived from primitive pluripotential cells, which have the potential for a wide range of differentiation.[11] As such, phenotypic heterogeneity is observed in different tumors as well as in the same tumor.

Approximately 100 cases of extra renal MERT have been reported, which demonstrated partial and global rhabdoid features on conventional microscopic examinations. Colonic MERT is an extremely rare tumor that has been described in elderly individuals. It is typically located proximal to the transverse colon. To our knowledge, only 10 cases of primary rhabdoid colonic tumors (RCT) have been reported, and our patient, at 31 years of age, is the youngest (Table 1).

The two described histologic types of RCT are pure and composite. Composite refers to adenocarcinoma with rhabdoid features and has been associated with polyposis syndromes. Three RCT out of the 10 previously reported tumors were composite and associated with multiple polyposis.

Treatment of colonic MRTs is typically surgical resection without chemotherapy or radiotherapy. Prognosis is very poor in cases with metastases. Given that the tumors are so rare, no clear consensus exists about the choice of chemotherapeutic agents. Single agent chemotherapy with bevacizumab, cetuximab[7] and multi-agent chemotherapy with capecitabin and oxaliplatin has been described.[2] Recent studies have suggested considering monoclonal antibodies against

Table1. Cases of Primary Rhabdoid Colonic Tumors

Authors	Age (years)	Gender	Site	Size (cm)	Histology	Outcome	Metastasis
Chetty and Bhathal[3]	72	Female	Cecum	6x5	Composite	Dead (3 months)	Liver, nodes
Yang et al.[6]	75	Male	Transverse colon	15x10	Pure rhabdoid	Dead (2 weeks)	Nodes
Marcus et al.[5]	84	Female	Transverse colon	7x6	Pure rhabdoid	Alive (12 months)	None
Nakamura et al.[1]	76	Male	Cecum	14x8	Pure rhabdoid	Dead (12 wks)	Liver, nodes
Kono et al.[4]	66	Male	Cecum	13x13	Composite	Dead (6 wks)	Peritoneum, nodes
Pancione et al.[7]	71	Male	Cecum	10x10	Pure rhabdoid	Dead (8 months)	Peritoneum, liver
Remo et al.[2]	73	Female	Cecum	10x8	Composite	Dead (6 months)	Node
Lee et al.[8]	62	Male	Sigmoid	4.5x4	Composite	Alive (36 months)	Node
Lee et al.[8]	83	Female	Rectum	6.5x4.3	Composite	Dead (1 month)	Liver, lung, nodes
Samalavicius et al.[9]	49	Male	Rectum	7	Composite	Dead (7 months)	Liver, nodes
Current case	***31***	***Female***	***Cecum***	***9x4***	***Pure rhabdoid***	***Dead (4 months)***	***Nodes, liver, bone***

the epidermal growth factor receptor for RCTs that exhibit "wild type" KRAS gene,[2,4,7] however no case reports were found where this was used.

In summary, rhabdoid tumors of the gastrointestinal tract are rare and associated with a poor prognosis. They do not respond to conventional therapeutic regimens. A better understanding of the genetic and molecular basis of these tumors may help guide management to improve prognosis. ■

References

1. Nakamura I, Nakano K, Nakayama K, et al. Malignant rhabdoid tumor of colon: report of a case. Surg Today. 1999;29:1083-1087.
2. Remo A, Zanella C, Molinari E, et al. Rhabdoid carcinoma of the colon: a distinct entity with a very aggressive behavior: a case report associated with a polyposis coli and review of the literature. Int J Surg Pathol. 2012;20:185-190.
3. Chetty R, Bhathal PS. Caecal adenocarcinoma with rhabdoid phenotype: an immunohistochemical and ultrastructural analysis. Virchows Arch A. 1993;422:179-182.
4. Kono T, Imai Y, Imura J, et al. Cecal adenocarcinoma with prominent rhabdoid feature: report of a case with immunohistochemical, ultrastructural, and molecular analyses. Int J Surg Pathol. 2007; 15:414-420.
5. Marcus VA, Viloria J, Owen D,et al. Malignant rhabdoid tumor of the colon: report of a case with molecular analysis. Dis Colon Rectum. 1996;39:1322-1326.
6. Yang AH, Chen WYK, Chiang H. Malignant rhabdoid tumour of the colon. Histopathology. 1994;24:89-91.
7. Pancione M, Di Blasi A, Sabatino L, et al. A novel case of rhabdoid colon carcinoma associated with a CpG island methylator phenotype and BRAF mutation. Hum Pathol. 2011;42:1047-1052.
8. Lee SH, Seol H et al. Rhabdoid colorectal carcinomas: reports of two cases. Korean J Pathol. 2013 Aug;47(4):372-7.
9. Samalavicius NE, Stulpinas R et al. Rhabdoid carcinoma of the rectum. Ann Coloproctol. 2013 Dec;29(6):252-5.
10. Weeks DA, Beckwith JB, Mierau GW, et al. Rhabdoid tumor of kidney. A report of 111 cases from the National Wilms' Tumor Study Pathology Center. Am J Surg Pathol. 1989;13:439-58.
11. Ota S, Crabbe DC, Tran TN, et al. Malignant rhabdoid tumor. A study with two established cell lines. Cancer 1993;71:2862-2872.