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Rosai-Dorfrman disease of the colon

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Rosai-Dorfman Disease of the Colon

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Rosai-Dorfman disease (RDD), formerly referred to as sinus histiocytosis with massive lymphadenopathy, was first described by Rosai and Dorfman in 1969 (1). Initially thought to be a disease limited to lymph nodes, RDD is now recognized to involve a variety of organs. Although gastrointestinal involvement has been reported, it remains rare. To our knowledge, there have been no cases of RDD reported in the gastroenterology literature and no cases evaluated with endoscopic ultrasound.

Case Report

A 50 year old African American female was found to have hemoccult positive stool and anemia. The patient described intermittent hematochezia for 6 months but reported no other gastrointestinal or systemic symptoms. Colonoscopy revealed numerous firm focally pigmented extramucosal nodules 5-25 cm from the anal verge (Figure A) with the remainder of the colon appearing normal. Endoscopic

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ultrasound demonstrated well-demarcated hypoechoic submucosal masses with intact muscularis propria (Figure B) and multiple oval peri-colonic lymph nodes. (Figure C). A snare biopsy of a representative nodule was performed.

Histologically, the nodule contained a chronic mixed inflammatory cell infiltrate, bands of fibrosis and large histiocytes with abundant pale eosinophilic cytoplasm and round vesicular nuclei. The histiocytes expanded both the lamina propria and the submucosa, the latter of which was massively expanded (Figure D). The histiocytes were immunoreactive for S100, CD68 and vimentin but negative for CD1a, alpha smooth muscle actin, c-Kit, synaptophysin, chromogranin, AIK-1, NSE, neurofilament protein and CD34. Emperipolesis with engulfment of plasma cells and neutrophils was most evident with the immunohistochemical stains for S100 and CD68.

Discussion

RDD is a rare histiocytic proliferative disorder of unknown etiology. Typically, it presents in children or young adults as painless cervical lymphadenopathy associated with fever, night sweats, malaise, and weight loss. There is a slight predilection for males and individuals of

African descent (2). Laboratory abnormalities may include an elevated erythrocyte sedimentation rate, anemia, neutrophilia, and polyclonal hypergammaglobulinemia. Extranodal involvement is reported to occur in 40% of the cases (2), most commonly involving bone, skin and soft tissue, central nervous system, eye and orbit, salivary glands and respiratory tract. Gastrointestinal tract involvement is rare, with a few case reports of disease involving the jejunum, appendix, colon, liver and pancreas (3). The majority of patients with gastrointestinal involvement have been asymptomatic with the disease being discovered incidentally. Lauwers (3) recognized a predilection of RDD to involve the more distal portions of the gastrointestinal tract with concomitant involvement of other nodal and/or extranodal sites.

Histologically, RDD is characterized by the presence of S100 positive and CD1a negative histiocytes, which exhibit emperipolesis or lymphocytophagocytosis, in the absence of eosinophils. The histiocytes are localized to the submucosa with occasional focal extension into the lamina propria. These features allow for distinction between RDD and Langerhans cell histiocytosis.

Although RDD is considered idiopathic, some evidence exists to suggest that immune dysfunction (4) and viral (EBV, HHV-6) infections (5, 6) play a role in its pathogenesis.

Two distinct disease patterns have been described. The more common pattern involves nodal disease from a single region and is typically self-limited with a tendency for total spontaneous regression. The second pattern, which includes disease with gastrointestinal involvement, involves several nodal or extranodal sites and usually runs a protracted course with intermittent exacerbations and remissions. Disease severity is dictated by the number and type of extranodal sites involved. Severe disease has been treated with chemotherapy and radiation therapy with variable success (7,8).

Thus far, our patient appears to have disease limited to her colon and peri-colonic lymph nodes. She remains asymptomatic and will be monitored for the development of disease related symptoms.

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References

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Figures Legends

Figure A: Endoscopic image of the sigmoid colon showing numerous extramucosal nodules.

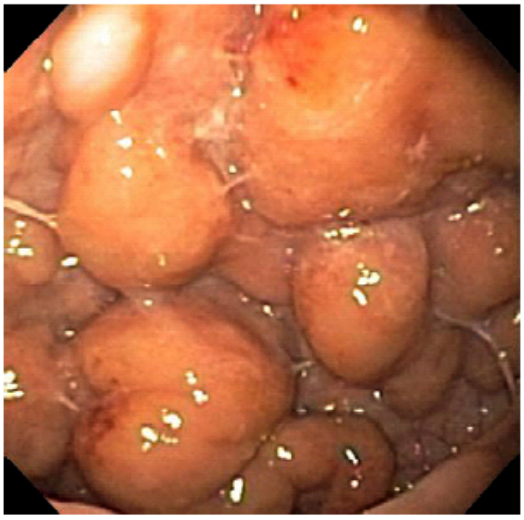


Figure B: Endoscopic ultrasound imaging of the sigmoid colon at 5 MHz demonstrating well-demarcated hypoechoic submucosal masses (white arrows) with intact muscularis propria (yellow arrow).

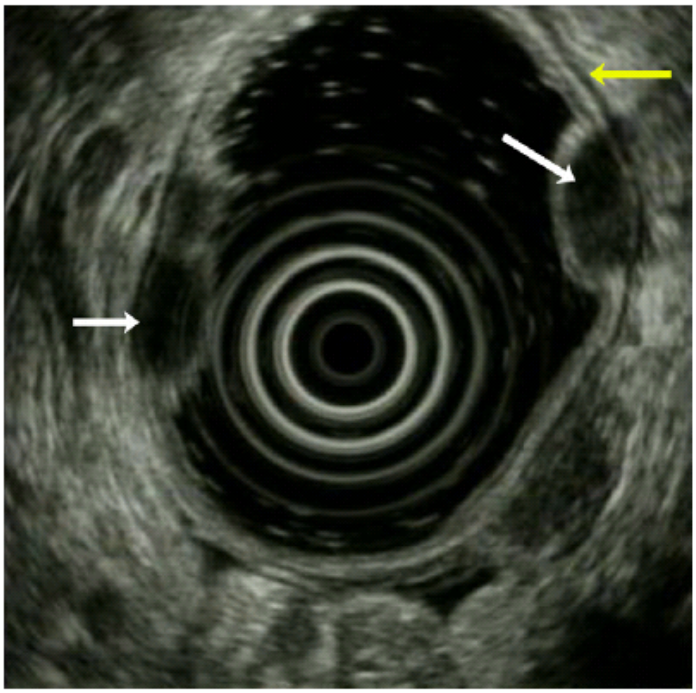


Figure C: Endoscopic ultrasound imaging of the sigmoid colon at 5 MHz demonstrating well-demarcated hypoechoic submucosal masses and peri-colonic lymph nodes (white arrows).

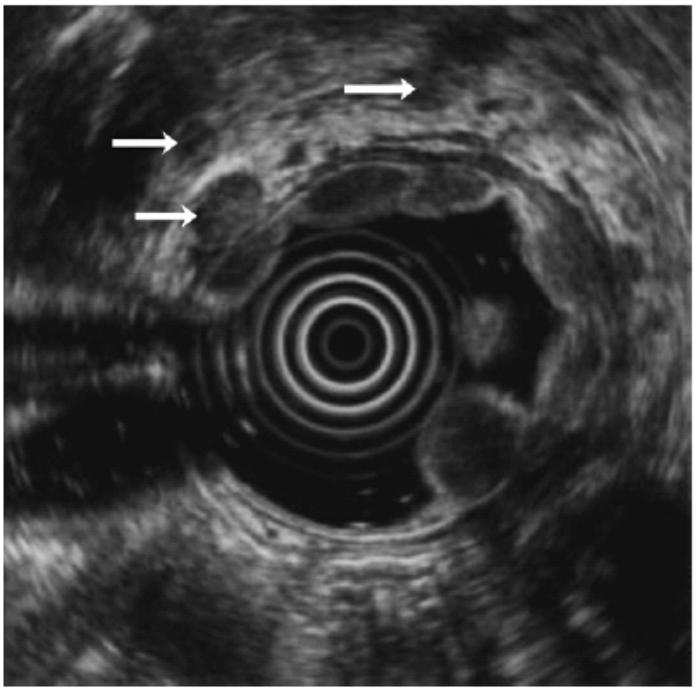
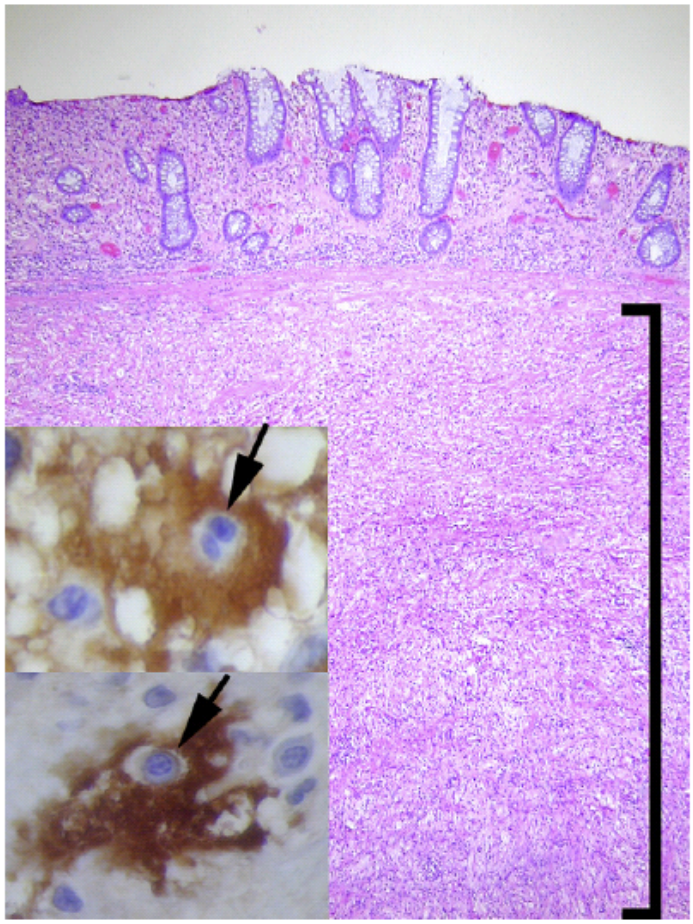


Figure D: Low magnification (H&E 40X original magnification) showing striking submucosal expansion (only part of which is shown here) marked by bracket. Insets, high magnification (600X original magnification) of S100 positive histiocytes with emperipolesis of neutrophil (upper, arrowed) and plasma cell (lower, arrowed).



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