Adult Hirschsprung Presenting As Chronic Constipation

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INTRODUCTION

Hirschsprung disease is caused by the absence of ganglion cells in the colon and most commonly diagnosed in the neonatal period. It typically affects 1 in 5000 live births and the most common symptoms are constipation, abdominal distension, failure to pass meconium, and vomiting during the first days of life. Adult Hirschsprung disease is a very uncommon diagnosis and is misdiagnosed as functional constipation.

Presentation

The patient is a 34-year-old female with history of chronic constipation since childhood who presented to the emergency department with abdominal pain, distension, nausea, and vomiting. Patient's family reported that she required an enema shortly after birth, multiple bowel regimens throughout her life without success, and multiple unremarkable colonoscopies. Most recently, she was started on Linzess which worsened her abdominal pain.

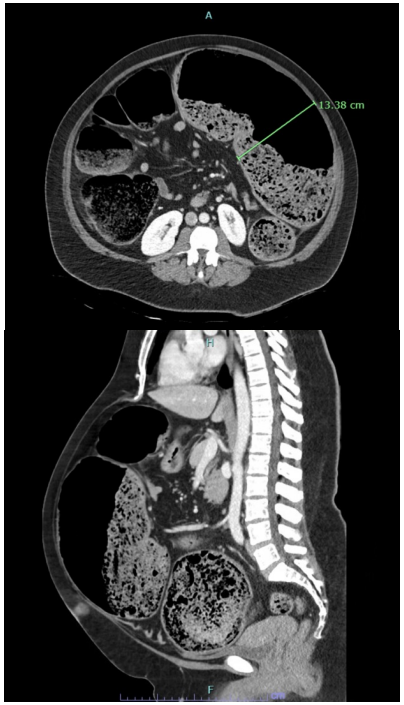

Figure 1: Admission CT showing distension of colon up to 14cm in the axial and sagittal planes

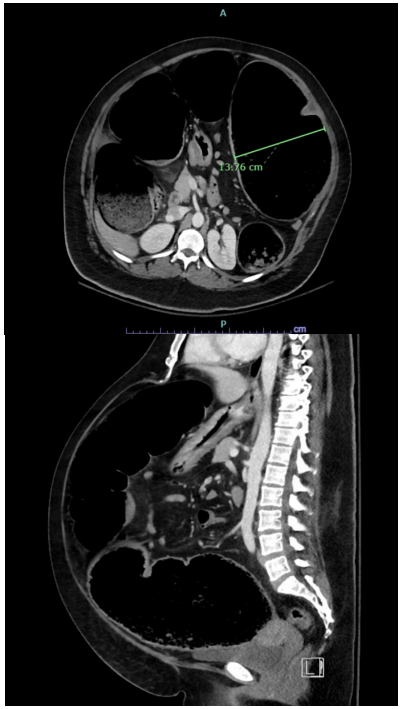

Figure 2: Pre-operative CT showing continued distension and new pneumatosis in the ascending colon

Clinical Course

After initial improvement with bowel regimen and resuscitation, the patient developed increasing abdominal pain and physical exam prompting repeat imaging demonstrating new found pneumatosis throughout the colon. She underwent an open total abdominal colectomy with end ileostomy and was found to have a necrotic right colon intra-operatively. Final pathology demonstrated a 2cm segment of rectosigmoid compatible with Hirschsprung disease.

CONCLUSIONS

Adult Hirschsprung disease is a relatively rare diagnosis and is more commonly written off as chronic constipation. In young adults with history of chronic colonic dysmotility, it is important to consider Adult Hirschsprung. In patient's who are appropriately diagnosed, the treatment is commonly surgical intervention with pull through surgery, allowing patient to have restoration of normal bowel function.

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