Title

An Adult with Megacolon — the Differential Diagnosis of Hirschsprung's Disease

Journal

Proceedings of UCLA Health, 20(1)

Authors

Berg, Christopher J.
Hernandez, Estebes A.

Publication Date

2016-11-03

CLINICAL VIGNETTE

An Adult with Megacolon – the Differential Diagnosis of Hirschsprung's Disease

Christopher J. Berg, M.S., and Estebes A. Hernandez, M.D.

Abstract

Hirschsprung's disease (HD), also known as congenital aganglionic megacolon, is an uncommon disease caused by a failure of migration of neural crest cells to the distal colon. While the vast majority of patients present as neonates, with bilious emesis, distension, and failure to pass meconium, at least 10% of patients will present after three years of age. We present an adult who had chronic constipation since infancy and who presented with megacolon and fecal impaction. We discuss the differential diagnosis of a patient evaluated for Hirschsprung's disease with emphasis on work-up and treatment.

History of present illness

A 36-year-old male with history of chronic constipation presented with abdominal pain and constipation. His constipation began at seven months of age when he transitioned to eating solid food. On average, he would have a bowel movement once every 1-2 months, where he would pass very hard stool with much straining. His symptoms would marginally improve with increased water intake, exercise, and a diet high in fruits and vegetables. Over the counter bowel regimens were not effective. He reported that his mother also suffers from a similar condition. He had never undergone colonoscopy or workup for constipation or otherwise sought medical care for his symptoms. He recalls requiring manual dis-impaction as a child.

He was in his usual state of health until four months prior to admission, where he noted worsening constipation with abdominal pain, distension, and bloating. He attributed the change in symptoms to a new job, which prevented adequate fluid intake. He reported having liquid stools intermittently since three months prior to examination, but no solid stool output for the last four months. He was passing flatus. He noted the pain was worse while standing and while supine but improved while lying on his left side. Increased water intake and a salt water enema did not relieve symptoms. Since the worsening constipation, he had reduced his oral intake to water and soups. He reported a 10kg weight loss over the last four months. He had not been febrile but reported having nausea without vomiting.

Physical Examination

On presentation to ED, the patient was afebrile with a heart rate of 137 beats per minute, blood pressure was 126/79 mmHg, and his oxygen saturation was 100% on room air. The patient appeared non-syndromic, although cachectic, ill, and in moderate pain. Abdominal exam was significant for a markedly distended abdomen with palpable colon with diffuse tenderness and hypoactive bowel sounds. Rectal examination revealed a large amount of hard stool palpable in the vault with gas and liquid passing around the stool. Physical exam was otherwise unremarkable. Labs on admission are shown in Table 1.

Clinical Course

A CT Abdomen Pelvis with contrast showed marked dilatation of the colon and rectum with a large fecal burden but otherwise without mass or evidence of pneumatosis or free air (Figure 1A). No clear transitional point was visualized. The patient was taken to the operating room under general anesthesia and placed in high lithotomy position where three "buckets" full of stool were manually dis-impacted and more stool was removed with saline irrigation.

After manual dis-impaction, the patient was able to have several large bowel movements with a bowel regimen of Golytely, daily mineral oil enemas, and miralax. Conventional radiograph taken two days following manual dis-impaction showed a significant decrease in the stool burden (Figure 1B). The patient was also started on total parenteral nutrition given his poor nutritional status. The patient was stable for discharge four days following manual dis-impaction at which the patient had lost 8 kg.

At a three-week follow-up, the patient reported daily stooling with intermittent miralax usage with improved oral tolerance and weight gain. The patient's abdomen was non-tender and non-distended on exam and laboratory values gathered at this time are shown in Table 1. At a follow-up appointment one month later, the patient reported having 1-2 bowel movements a week with full appetite and continued weight gain. The most likely diagnosis was thought to be congenital aganglionic megacolon (Hirschsprung's disease). He was scheduled for further outpatient evaluation including anorectal manometry, rectal suction biopsy, and sigmoidoscopy; unfortunately, he was lost to follow-up.

Discussion

Hirschsprung's disease (HD) is a relatively rare disease of the absence of ganglion cells in the submucosal and myenteric neural plexuses of the distal hindgut, resulting in loss of peristalsis in the affected region and subsequent functional obstruction.[1–3] The pathogenesis of the disease is a failed cephalocaudal migration of neural crest cells along the neural tube to the rectum. The severity of disease corresponds to the length of aganglionic colon.[2] Incidence is approximately 1 in every 5,000 live births with a 3:1 male predominance. The overwhelming majority of these patients are diagnosed as children but at least 10% of patients present after the age of three.[1] While patients diagnosed as infants generally present with bowel obstruction, the vast majority of patients diagnosed as adults report lifelong constipation requiring laxative or enema therapy, three quarters of them experiencing symptoms at three years of age or younger.[4]

The severity and extent of disease in adults diagnosed with HD is significantly less than those diagnosed as infants or children. A meta-analysis of Hirschsprung's disease in adults revealed that the majority of patients had disease restricted to the rectum or sigmoid colon, resulting in less severe symptoms. Adult cases involving the descending, transverse, or entire colon were extremely rare.[4,5]

The gold standard for diagnosis is full thickness biopsy of the rectum but it is inconvenient and invasive, requiring anesthesia. In comparison rectal suction biopsy has been shown to be equally sensitive and specific for diagnosing HD and can be performed as an office procedure. Histology reveals absence of ganglion cells in the submucosa as well as increased acetylcholinestase activity.[6] Other corroborating evidence of HD include absent recto-anal inhibitory reflex (RAIR) on contrast enema and anorectal manometry.[5]

Once the diagnosis is made, the primary modality of therapy for Hirschsprung's disease is surgical, although less severe cases can be managed with medical therapy. In the infant, treatment often includes decompression of the colon, resection of the aganglionic section of colon, and either primary pull-through anastomosis or colectomy to bridge to pull-through procedure (in the setting of enterocolitis or prohibitively dilated colon. There are a variety of pull-through techniques, that are performed as per the surgeon's preference.[7] Although there is no compelling evidence to suggest that a certain technique offers superior outcomes, the modified Duhamel procedure is the most commonly performed operation.[4,5,8] Similar outcomes have been reported for laparoscopic and transanal approaches.[9]

Differential Diagnosis

In the patient with symptoms of HD, but with ganglion cells on rectal biopsy, several other diagnoses must be considered, including hypoganglionosis, intestinal neuronal dysplasia (IND), internal anal sphincter achalasia (IASA), and megacystis microcolon intestinal hypoperistalsis (MMIHS).[10] Each of these disease present very similarly to HD, although preferred surgery may different.

Hypogangliosis is a very rare disease of decreased ganglion cells in the colonic submucosa. While there are important histological differences between hypoganglionosis and HD, including decreased numbers of nerve cells in the plexus area, and acetylcholinesterase staining in the affected bowel specimens, the presentation and treatment are very similar.[10]

IND has two distinct subtypes with separate clinical presentations. Type A is less common (<5%) and more severe, presenting with intestinal obstruction, hematochezia and diarrhea. In contrast, Type B IND present much more similarly to HD, with abdominal distention and constipation. The diagnostic criteria are controversial, although hyperganglionosis is an important component. Accepted treatment includes initial conservative treatment with laxatives and enemas, followed by surgical options, if symptoms persist. Internal sphincter myomectomy and botulinum toxin injection have been reported, but resection and pull-through operations, the mainstay of HD therapy, are only indicated in the most severe cases.[10,11]

The diagnosis of IASA requires absence of rectosphincteric reflex on anorectal manometry and presence of ganglion cells on rectal biopsy along with symptoms similar to HD. Similarly to IND, treatment of IASA has been successful with posterior internal anal sphincter myectomy and botulinum toxin injection.[10]

MMIHS is an extremely rare congenital condition involving a diminutive colon and enlarged bladder with the symptoms of intestinal obstruction, inability to void, and abdominal distention. The etiology of MMIHS is still unclear, although it appears to have a female predominance and possible familial inheritance. Diagnosis includes dilated bladder and hydronephrosis on prenatal ultrasound, as well as a dire clinical presentation. Reported surgical interventions involve multiple organs, often resulting in TPN requirements and possible visceral organ transplants. While these patients have done very poorly historically, improvements in transplantation and TPN have resulted in better outcomes in the last decade.[10]

Conclusion

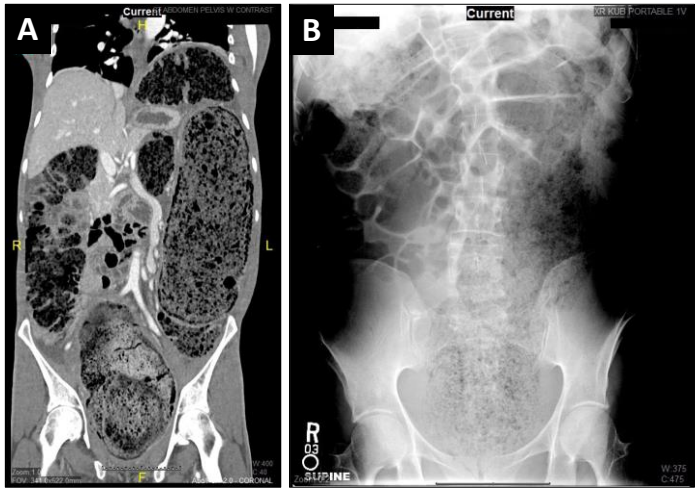
We presented an adult with symptoms of constipation since infancy who was admitted with megacolon and fecal impaction. Although the patient's clinical presentation and imaging were consistent with HD, we discuss the diagnosis and treatment of Hirschsprung's disease, as well as other diagnoses that must be considered until rectal biopsy proves absence of ganglionic cells in the affected colon, thus securing the diagnosis of HD.

Figures and Tables

Table 1: Laboratory Values at admission and at 3-week follow-up

Laboratory Measurement	**Value at admission**	**Value at 3-week follow-up**
Na (mEq/L)	133	141
K (mEq/L)	3.8	4.4
Cl (mEq/L)	94	100
HCO3 (mEq/L)	24	26
BUN (mg/dL)	10	16
Cr (mg/dL)	0.6	0.7
Glucose (mg/dL)	128	92
Ca (mg/dL)	8.2	9.4
Mg (mEq/L)	1.7	
Phos (mg/dL)	4.2	
AST (U/L)	13	16
ALT (U/L)	15	26
Alk Phos (U/L)	76	74
Bilirubin, total (mg/dL)	0.5	0.5
Amylase (U/L)	42	
Lipase (U/L)	11	
Albumin (g/dL)	3.1	4.6
Prealbumin (mg/dL)	8.7	
Total Protein (g/dL)	5.9	7.0
ESR (mm/hr)	19	
CRP (mg/L)	4.6	
WBC ($x10^3/\mu L$)	16.37	
Hemoglobin (g/dL)	13.3	
Hematocrit (%)	38.2	
MCV (fL)	89.0	
Plt count ($x10^3/\mu L$)	431	
Neutrophils (%)	80.8	
Lymphocytes (%)	11.5	
Monocytes (%)	7.4	
Eosinophils (%)	0.1	
Basophils (%)	0.2	
TSH ($\mu U/mL$)	4.7	

Figure 1: A) Computed tomography with contrast of the abdomen and pelvis prior to manual dis-impaction, showing large stool burden. No definite transition point was visualized. B) Conventional radiography of abdomen and pelvis after manual dis-impaction. A large amount of stool is still visualized in the rectum and descending colon.



REFERENCES

1. **Arshad A, Powell C, Tighe MP.** Hirschsprung's disease. *BMJ*. 2012 Oct 1;345:e5521. doi: 10.1136/bmj.e5521. PubMed PMID: 23028095.
2. **Parisi MA, Kapur RP.** Genetics of Hirschsprung disease. *Curr Opin Pediatr*. 2000 Dec;12(6):610-7. Review. PubMed PMID: 11106284.
3. **Kenny SE, Tam PK, Garcia-Barcelo M.** Hirschsprung's disease. *Semin Pediatr Surg.* 2010 Aug;19(3):194-200. doi: 10.1053/j.sempedsurg.2010.03.004. Review. PubMed PMID: 20610192.
4. **Vorobyov GI, Achkasov SI, Biryukov OM.** Clinical features' diagnostics and treatment of Hirschsprung's disease in adults. *Colorectal Dis*. 2010 Dec;12(12):1242-8. doi: 10.1111/j.1463-1318.2009.02031.x. PubMed PMID: 19674017.
5. **Doodnath R, Puri P.** A systematic review and meta-analysis of Hirschsprung's disease presenting after childhood. *Pediatr Surg Int.* 2010 Nov;26(11):1107-10. doi: 10.1007/s00383-010-2694-2. Review. PubMed PMID: 20725836.
6. **De Lorijn F, Reitsma JB, Voskuijl WP, Aronson DC, Ten Kate FJ, Smets AM, Taminiau JA, Benninga MA.** Diagnosis of Hirschsprung's disease: a prospective, comparative accuracy study of common tests. *J Pediatr*. 2005 Jun;146(6):787-92. PubMed PMID: 15973319.
7. **Kessmann J.** Hirschsprung's disease: diagnosis and management. *Am Fam Physician*. 2006 Oct 15;74(8):1319-22. Review. PubMed PMID: 17087425.
8. **Barnes PR, Lennard-Jones JE, Hawley PR, Todd IP.** Hirschsprung's disease and idiopathic megacolon in adults and adolescents. *Gut*. 1986 May;27(5):534-41. PubMed PMID: 3699562; PubMed Central PMCID: PMC1433503.
9. **Thomson D, Allin B, Long AM, Bradnock T, Walker G, Knight M.** Laparoscopic assistance for primary transanal pull-through in Hirschsprung's disease: a systematic review and meta-analysis. *BMJ* Open. 2015 Mar 24;5(3):e006063. doi:10.1136/bmjopen-2014-

006063. Review. PubMed PMID: 25805527; PubMed Central PMCID: PMC4386272.
10. **Puri P, Gosemann JH.** Variants of Hirschsprung disease. *Semin Pediatr Surg*. 2012 Nov;21(4):310-8. doi: 10.1053/j.sempedsurg.2012.07.005. Review. PubMed PMID:22985836.
11. **Puri P.** Intestinal neuronal dysplasia. *Semin Pediatr Surg*. 2003 Nov;12(4):259-64. Review. PubMed PMID: 14655165.

Submitted November 3, 2016