

Congenital aganglionic megacolon in Nigerian adults: Two case reports and review of the literature

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Abstract

Congenital aganglionic mega colon (Hirschsprung's disease) is a motor disorder in the gut, due to a defect in the craniocaudal migration of the neuroblast originating from the neural crest that occurs during the first twelve weeks of gestation, causing a functional intestinal obstruction, with its attendant complications, in infants. Despite modern pediatric practice, with emphasis on early diagnosis, Hirschsprung's disease is seen in adults in regions where perinatal care is limited. We report two cases of Nigerian adults with longstanding, recurrent constipation, getting relieved by laxatives and herbal enemata, and then presented to our Emergency Department with a history of progressive abdominal distention, colicky pain, occasional vomiting, and weight loss. Per rectal examination revealed a gripping sensation in the rectum, 10 cm from the anal verge, with rectal fecal load. Barium enema showed a grossly distended proximal large colon, with high fecal retention, with the transition zone at the middle one-third of the rectum. Due to difficulty in bowel preparation of these patients, emergency laparotomy was done. The first case had a diverting sigmoid colostomy and later had a low anterior resection. The second case had a one-stage procedure. Histology of both the cases showed aganglionosis of the stenotic segment and a normal distal rectum. Both patients had complete resolution of the symptoms, without complications, in a three-year follow-up. The related literatures were reviewed. Hirschsprung's disease should be considered in adults patient presenting with chronic constipation. Low anterior resection of the rectum would be a surgical option for the treatment of short and zonal segment of adult Hirschsprung's disease.

Key words: Adult, congenital, Hirschsprung's disease, mega colon

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Introduction

Hirschsprung's disease is the malformation of the hindgut characterized by the absence of intramural ganglion cells in the Meissner's and myenteric plexuses, manifested by the megacolon, leading to functional obstruction and colonic dilatation proximal to the affected segment.^[1,2] It occurs in 1 of 5000 live births, with an overall male: female ratio of 3:1 to 4:1, but when the entire colon is involved, the gender ratio approaches 1:1.^[3-5] The most common accepted theory of the etiology of Hirschsprung's disease is that there is a defect in the craniocaudal migration of neuroblasts originating from the neural crest, and recent studies show an association of Hirschsprung's disease with some chromosomal abnormalities and syndromes.^[6-12] The presentation of Hirschsprung's

disease is not common in adults because it is a congenital anomaly, and therefore, most often presents early in life, for surgical intervention.^[7,13,14] We report two cases in Nigerian adults with review of the literature.

Case Reports

Case 1

A 27-year-old man presented with recurrent constipation and abdominal distention since birth. The symptoms were relieved by enema. The constipation improved over the years

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until the last three years when he began to have recurrent constipation associated with abdominal distention, colicky pain, and occasional vomiting and weight loss. There was no hematochezia, no history of a previous surgery or family history. He had a herbal enema a week prior to presentation. On clinical examination, the abdomen was grossly distended with visible peristalsis, but there was no palpable mass. Digital rectal examination revealed a good anal sphincteric tone with gripping sensation 10 cm from the anal verge and high rectal fecal load, and brownish mucoid feces gushed out following the digital examination. A clinical diagnosis of Adult Hirschsprung's disease was made. Routine laboratory evaluation, including a complete blood count, urinalysis, and blood chemistry studies, were within normal limits. The barium enema showed a grossly dilated, large colon, with a transition zone at the middle one-third of the rectum. On account of the grossly dilated large colon with high fecal retention and difficulty in preparing the bowel, emergency laparotomy with diverting sigmoid colostomy was done. Twelve weeks post colostomy the patient had low anterior resection with colo-anal anastomosis. The resected segment was 25 cm in length. The histology showed aganglionosis of the narrowed segment, but a normal distal one-third rectum. The patient followed up for two years without constipation or other complications.

Case 2

A 30-year-old female presented in 2006, with recurrent episodes of constipation and abdominal distention, from the age of two years. The symptoms were relieved by the use of herbal enemas or liquid paraffin. She had two exploratory laparotomies in 1995 and 2000, at a General Hospital, without improvement. The last episode occurred four days prior to presentation, with constipation and progressive abdominal distention. There was no diarrhea, melena stool, or rectal bleeding. Clinically she was not in distress, the abdomen was grossly distended. Digital rectal examination revealed a normal anal sphincter, with a stenosed gripping segment of the rectum, about 10cm from the anal verge. Large brownish fluid fecal volume gushed out following the digital examination. A routine laboratory evaluation, including a complete blood count, urinalysis, and blood chemistry studies, was normal. The barium enema studies showed marked dilatation of the sigmoid colon, filled with pellets of feces and the transition zone at the lower rectal segment [Figure 1], which was suggestive of Hirschsprung's disease. At laparotomy, the sigmoid and the rectum proximal to the stenosed distal rectum were grossly dilated, hypertrophied, and filled with hard pellet-like feces. Low anterior resection was done. The segment of the dilated sigmoid and the rectum, including the stenosed region, were resected [Figure 2] and the histology of the stenosed segment revealed rudimentary aganglionic cells consistent with Hirschsprung's disease. The patient was followed-up for three 3years. The defecation was satisfactory and without complications.



Figure 1: Photograph showing segment of the resected megacolon filled with faeces



Figure 2: Barium enema showing rectum full of faeces and transition zone

Discussion

Hirschsprung's Disease (HD) is characteristically manifested at birth and diagnosis is made in the neonatal period, in a majority of cases.^[7,13,15-18]

The most common accepted theory of the etiology of Hirschsprung's disease is that there is a defect in the craniocaudal migration of neuroblasts originating from the neural crest, which occurs during the first 12 weeks of gestation, and other the mechanisms proposed include defects in the differentiation of neuroblasts in the ganglion cells and accelerated ganglion cell destruction in the intestine^[6,7] More than eight genetic mutations have been identified with the disease.^[8,9] The RET proto-oncogene is most affected and accounts for 50% of the familial and 20% of the sporadic cases.^[8,9] Certain RET proto-oncogene polymorphisms are associated with a phenotype of short or long segment Hirschsprung's disease.^[10] Hirschsprung's disease is associated with other chromosomal abnormalities and syndromes such as trisomy 21, cardiac diseases,

congenital hypoventilation syndrome, MEN2, Waardenburg syndrome, and the Smith-Opitz syndrome.^[6,11,12]

Hirschsprung's disease in the adult is about 5% and is frequently misdiagnosed as chronic constipation.^[14,18,19] The presentation of the disease is in the early life when immediate diagnosis and prompt surgical intervention is carried out. Those that present late in life as in these two cases warrant a proper medical history and evaluation, to exclude other causes of chronic constipation, such as, idiopathic megacolon and chagas disease.^[7] The late presentation of Hirschsprung's disease is very rare in advanced countries.^[13,20,21] However it is still common in our environment, probably because of lack of adequate perinatal care in our rural setting, where a majority of the populace live.^[22] Approximately 75-80% of the patients with Hirschsprung's disease have a transition zone in the rectosigmoid area, but 2-3% of Hirschsprung disease patients have the so-called 'ultra-short segment' or zonal segment Hirschsprung's disease.^[7,23] The existence of a single or double zonal or ultra-short Hirschsprung's diseases are often questioned, but studies done by Cornelis *et al.*, have confirmed the existence of such diseases.^[24,25] The skip area of the normal colon can be attributed to an extramural phase of neuroblast migration, which is unique to the colon.^[26] The two index cases present very early in life, with longstanding constipation, which is relieved by laxatives and herbal enemas. Perhaps that is why the parents never bother to seek medical advice. Again the two cases reside in the rural area, where probably specialized medical personnel may not be present to properly assess them.

The second case had two laparotomies, which unfortunately did not solve her clinical problem. This was probably due to the atypical clinical features at presentation and a poor index of suspicion. At this age, patients usually present with complications like obstructive colitis, sigmoid volvulus, or sub-acute intestinal obstructions.^[18] The index cases had features of obstruction and enterocolitis. There were no other obvious, associated congenital anomalies or syndromes in these cases. However, we had no facilities in our setting to ascertain the presence or absence of genetic or chromosomal anomalies.^[8,9,10] Chen *et al.*, reported a 26-year-old lady who presented with severe intestinal obstruction that was refractory to conservative management, and she later had a one-stage procedure.^[14]

Case 1 had colostomy initially in order to decompress the grossly distended abdomen and that facilitated significant reduction in the size of the colon, and rectosigmoidectomy with coloanal anastomosis was carried out without creating temporary protecting colostomy.

Case 2 had a one-stage approach. The choice of the operative approach depends on identifying the distal most

segment of the bowel with ganglion cells, which is frequently suggested by the contrast enema and then confirmed by biopsy with frozen section examination, accomplished in the operating room^[13], but due to lack of facilities, we adopted less than ideal techniques for the management of Hirschsprung's disease.^[7,13,15] Technical approaches different from Swenson's original pull-through procedure have been developed and further modified to the present development of the laparoscopic approach, for the surgical treatment of Hirschsprung's disease.^[1,3,16,18,19,27-29] Both patients had short or zonal segment of aganglionosis close to the anal canal, hence, low anterior resection (state) was done.^[3,16,22] A follow-up after two years showed an excellent functional result after the surgery.

Due to rapid recent advances made in understanding the genetics and molecular pathology of Hirschsprung's disease, stem cell-based therapy for Hirschsprung's disease is being advocated.^[10,24,29,30]

Conclusions

In adults, although there are other causes of mega colon, when they present with chronic constipation dates from new-born, Hirschsprung's disease should be considered. Low anterior resection would be a perfect surgical option for the treatment of short or zonal segment adult Hirschsprung's disease.

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