Adulthood hirschprung's disease : a report of 4 cases in Ile – Ife. Nigeria

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Abstract

Background: Hirschsprung's disease in adulthood is very rare and is often misdiagnosed. We present four cases of adulthood Hirschsprung's disease seen in the last two decades to illustrate challenges accompanying its diagnosis and management.

Method : This descriptive case series included cases of histologically proven Hirschsprung's seen in adulthood at the Obafemi Awolowo University Teaching Hospitals Complex in the last two decades (1991 – 2011). The clinical data, radiological investigations, details of surgical treatment, histological diagnosis, outcomes and complications were analyzed.

Result : There were 4 adult patients, 3 males and 1 female with age ranging from 17 to 74 years (mean 23 years). Each patient presented with sub acute intestinal obstruction needing two staged procedures of initial colostomy followed by definitive procedure of low anterior resection (State procedure) in 3 patients and Swenson – Bill procedure in one. There was one mortality and good long term outcome in the remaining three.

Conclusion : This review presented the oldest patient presenting with adult Hirschsprung's and the highest mean age of any case series. Four patients with adulthood Hirschsprung's disease managed by two operative procedures enabled comparison of operative outcome with respect to complications and functional outcomes. Mortality seems to correlate with presentation at old age, which is usually due to life long self-management of chronic constipation. Though very rare, a high index of suspicion of adulthood Hirschprung's disease should be maintained in adult patients with recurrent chronic constipation needing lifelong laxative, enema or mechanical wash-out.

Keywords : *Adulthood Hirschprung disease, Megacolon, Nigeria*

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Résumé

Contexte: Hirschsprung la maladie à l'âge adulte est très rare et est souvent mal diagnostiquées. Nous présentons quatre cas de l'âge adulte Hirschsprung la maladie vu au cours des deux dernières décennies pour illustrent les défis accompagnant son diagnostic et de la gestion.

Méthode : Ce descriptif série cas comprenaient les cas de prouvée histologiquement Hirschsprung a vu dans l'âge adulte à l'Université Obafemi Awolowo hôpitaux d'enseignement complexe au cours des deux dernières décennies (1991 - 2011). Les données cliniques, radiologiques les enquêtes, les détails du traitement chirurgical, diagnostic histologique, les résultats et les complications ont été analysées.

Résultat : Il y a eu 4 patients adultes, 3 hommes et 1 femmes d'âge allant de 17 à 74 ans (moyenne 23 ans), chaque patient s'est présenté avec sous occlusion intestinale aiguë nécessitant deux étapes de procédures initiales de colostomie suivie de procédure définitive de faible résection antérieure (état procédure) chez 3 patients et Swenson - Bill procedure dans un. Il y a une mortalité et un bon résultat à long terme dans les trois autres.

Conclusion :, cet examen a présenté le plus ancien patient présentant des adultes Hirschsprung et de la plus haute moyenne d'âge est de toute façon série. quatre patients à l'âge adulte Hirschsprung la maladie gérés par deux procédures du dispositif activé comparaison des résultats du dispositif en ce qui concerne les complications et les résultats fonctionnels. Mortalité semble être en corrélation avec la présentation à la vieillesse, qui est généralement due à la vie longue l'auto-gestion de constipation chronique. Bien que très rares, un indice élevé de suspicion de l'âge adulte précoces de la maladie doit être maintenue chez les patients adultes avec récurrente constipation chronique nécessitant l'éducation laxatif, lavement ou mécanique wash-out.

Introduction

Hirschsprung's disease (HD) is a congenital abnormality of the ganglion cells in the bowel, it is characterised by absence of parasympathetic ganglia cells normally found in the intermuscular and submucosal nerve plexus of the bowel wall in the

distal bowel, but there is excessive amount of nerve fibrils and acetycholinesterase activity is also increased. It is commonly seen in newborns infants but could be rarely encountered in adulthood [1,2]. The term "Adult Hirschsprung's Disease" is usually employed when the diagnosis is established after the age of 10 years [3]. In 1964, Swenson observed that few patients lived beyond 12 to 14 years when their disease was not treated surgically [4].

Persistence of HD into adulthood is often misdiagnosed maybe because of rarity of this condition in adolescent and adulthood. Some patients may have milder disease and the condition may go undiagnosed because the proximal innervated colon can be hypertrophied and thus compensate for the distal aganglionic rectum. In those patients, the diagnosis may be first established during adulthood. Many patients usually give history of taking cathartics, using frequent enemas and progressive constipation or even life-long refractory constipation [2,5]. Most cases in adulthood have been diagnosed and treated in the adolescent and early adulthood stage [2,5], and it is indeed very rare for patients to carry Hirschsprung's disease to old age. A Search of English Medical literature particularly Medline in the past 50 years yielded very few reports of Hirschsprung disease in adulthood and elderly patients. We reviewed our experience and herewith present illustrative cases to highlight the challenges of diagnosis and management including surgical treatment and the long term outcome.

Patients and methods

Patient 1

A 74 year old retired hospital driver presented on 5th September 1991 with a year history of recurrent constipation characterised by passage of pellet like hard stool, two months history of progressive abdominal distension and colicky central abdominal pain of three days duration. He had been having recurrent episodes of milder constipation since childhood which resolved with the use of laxative and had not experienced any severe constipation like the one that caused him to present in the hospital. Physical examination revealed an acutely ill looking elderly man in painful distress, dehydrated, restless, tachycardic (pulse rate of 120/minute) and hypertensive (blood pressure of 180/90 mmHg). The abdomen was grossly distended with tympanitic percussion notes but had no palpably enlarged intra -abdominal mass, visible peristalsis or demonstrable ascites. The rectal examination revealed an increased anal sphincteric tone but no rectal mass and the prostate gland was not enlarged. Plain abdominal X-ray revealed dilated large bowel with faecal shadows,mottling and calcification. Haematological parameters and electrolytes were within normal limits. The initial clinical diagnosis was large bowel obstruction, secondary to a colonic tumour. For this he had emergency exploratory laparotomy. Intraoperative findings included a grossly distended large bowel commencing from the sigmoid colon up to the transverse colon in addition there was tapering of the distended sigmoid distally toward the presumably hypoplastic proximal rectum. No organic cause of obstruction was found. Other intrabdominal organs were grossly normal. The patient then had sigmoid loop colostomy. He later had flexible sigmoidoscopy and colonic biopsy. The histology of this was reported as colonic aganglionosis (Hirschsprung's disease). Eight months later he had colostomy closure and low anterior resection with colorectal anastomosis done. His unstable heamodynamic status intraoperatively precipitated acute renal tubular necrosis and subsequent acute renal failure which a session of haemodialysis could not ameliorate before finally succumbing on 4th day post surgery.

Patient 2

A 24 year old female student nurse presented on 3rd of December 2004 with an 8 years history of altered bowel habit characterized by recurrent episodes of alternating constipation and diarrhoea, passage of small hard pellet like stool and tenesmus. She developed recurrent abdominal distension over the 5 years prior to presentation which usually responded to use of cathartics and enema saponis. However the abdominal distension became worse and progressive 6 weeks prior to presentation. There was no family history of a similar condition. Examination essentially revealed a chronically ill looking depressed young lady with moderate weight loss and dehydration. The abdomen was grossly distended with tympanitic percussion notes but was not tender. Impacted solid faecal matter was felt on digital rectal examination.The initial clinical diagnosis was idiopathic acquired megacolon. She was resuscitated with antibiotics, intravenous fluid infusion. The blood chemistry was within normal limits. An emergency exploratory laparotomy was carried out and she had sigmoid loop colostomy and full thickness rectal biopsy. The operative findings included a grossly dilated sigmoid colon that was 48 centimeters in diameter with thickened wall, associated grossly dilated descending colon with

diameter of 24 centimeters all loaded with faeces. The rectum was normal in calibre and the mucosa was normal. There was no rectal mass except for faecaloma found at surgery. The post operative period was unremarkable. The histopathology of the full thickness rectal biopsy was reported as aganglionosis of the colon (Hirschsprung's disease). She had low anterior rectal resection at 9 months post presentation and was discharged home 2 weeks later. She has been followed up regularly in the surgical outpatient department since then and there has been no recurrence of chronic constipation and abdominal distension. She had gained weight and her bowel habit has normalized to bowel opening of once in a day or two.

Patient 3

A 25 year old secondary school leaver was first seen on 16th September 2009 with 4 weeks history of chronic constipation and abdominal distension without history of vomiting. He had been having recurrent episode of constipation since childhood but none of the episodes lasted more than a week. He had always taken cathartics to relieve chronic constipation. Neonatal history of non passage of meconium could not be ascertained. Physical eexamination revealed a distended abdomen which was tympanitic but with no mass or tenderness. Our initial diagnosis was colonic obstruction from a benign cause which was then uncertain. Plain x ray of the abdomen showed dilated large and small bowel. Abdominal computerized axial tomography scan (figures 1and 2) showed dilated large bowel with huge faecal matter and no organic cause of obstruction.

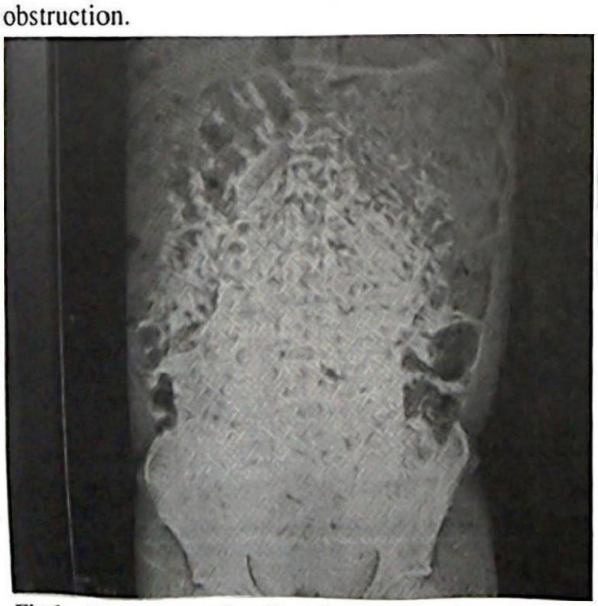

Fig.1: Scanogram of patient 3 pre operative showing large bowel laden with feaces

The sigmoid colon was redundantly dilated more than other large bowel reaching up to the diaphragm. He had exploratory laparotomy, sigmoidectomy and Devine colostomy. Histology of both end of the resected sigmoid colon loop revealed aganglionosis with hypertrophied and disorganised nerve fibres of the distal end of the resected sigmoid. One and half years later he had low anterior resection and closure of colostomy. The procedure was well tolerated and post operative period was uneventful. Outpatient clinic follow up has so far revealed no recurrent episode of chronic constipation. He now moves his bowel regularly once in a day as opposed to the initial bowel opening frequency of once in two to three weeks.

Patient 4

A 17 year old secondary school leaver presented as an emergency on 16th of October 2010 with a day history of sudden onset colicky abdominal pain with progressive abdominal distension. There was no associated vomiting and no previous history of constipation or similar episode of abdominal pain. He had orchidopexy for undescended testes about two years earlier.

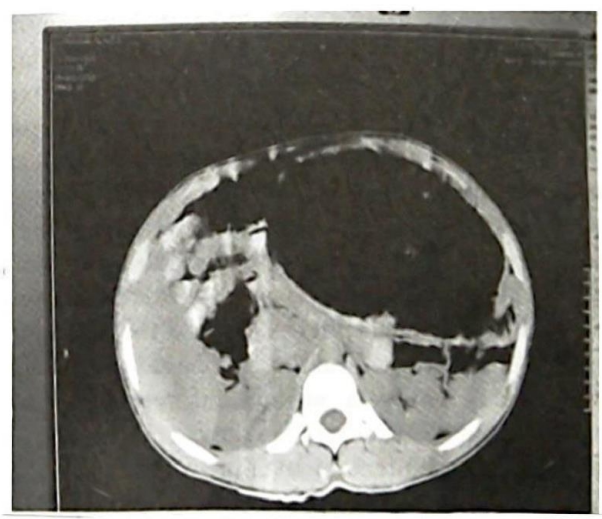

Fig. 2: CAT Scan of patient 3 coronal view showing grossly dilated sigmoid colon

Examination revealed an acutely ill looking young boy in painful distress with mild dehydration. The abdomen was generally distended, tense and tender. Bowel sounds were hypoactive. Plain abdominal X- ray showed dilated large and small bowel. Initial clinical diagnosis was large bowel obstruction possibly caused by congenital adhesive band. After fluid and electrolyte resucitation he had emergency exploratory laparotomy in which was found 150 mls of serous ascitic fluid, grossly

distended large bowel terminating at the rectosigmoid junction, multiple enlarged mesenteric lymph nodes. A sigmoid colostomy was carried out and a full thickness rectal biopsy taken and sent for histopathology confirmed Hirschsprung's disease (Figure 3). He subsequently had Swenson abdominoperineal pull through six months later.

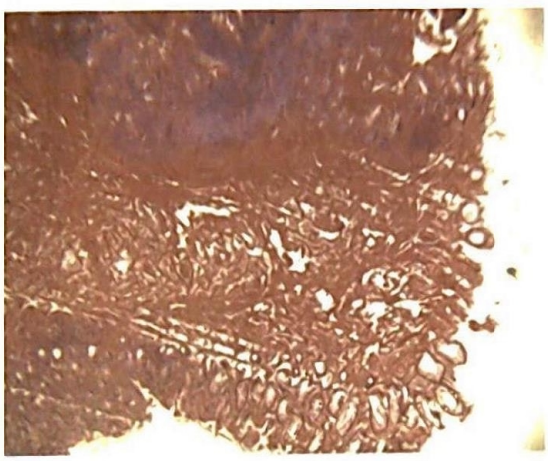

Fig.3: Sections show colonic tissue with absent ganglion cells in the muscularis mucosae and muscularis propria

Discussion

Adult Hirschsprung's disease is an uncommon disorder, since congenital aganglionosis is most often diagnosed and treated in neonatal period, infancy or early adulthood. The first well documented case of adult Hirschsprung's disease in English literature was reported by Rosin (1950) [6] in a 54 year old man and over the second half of the last century various reports of the condition has been made : Kempton (1954) [7] and Magliette (1960) [8] . This case series of adulthood Hirschsprung's disease includes one adolescent, two young adults and one elderly patient that were managed in our hospital over a 19 year period from 1991 to 2010. In the overall review of surgical English literature over the past 50 years the oldest patient so far was a 73 year old man reported by Elliot *et al* (1985) [9] . He was a year younger than the oldest patient in this series. The average age of 24.1 years [10] is being quoted for adult Hirschsprung disease in the literature in the past half a century. It is noteworthy that the average age at presentation in this series was 35 years, being a decade later than the average age in medical literature. Although the number of patients in the series was four, it could be said however that delay in presentation was a factor. Neglect and delayed presentation are due to patient's ignorance, poverty and reliance on self-care of chronic constipation and also failure of discovery of the condition at birth due to poor health delivery system of our country. It was the de-compensation of the proximal normal ganglionated segment of the large bowel resulting in acute intestinal obstruction that brought three of the patients to the hospital. The acute emergency clinical state therefore justified the use of colostomy as the first step in surgical therapy to decompress the decompensated bowel. Late presentation of Hirschsprung disease after 1 year of age has been recognized to be common in low resources countries such as Nigeria [11-13]. However there has been no case series of adulthood Hirschsprung disease such as this from Nigeria. This has by no means suggested the condition is not uncommon in Nigeria, misdiagnosis and underreporting characterizing the condition in a developing country may be responsible. To the best of authors knowledge the only report from Nigeria similar to the current series was in a 32-year old man [14]. The other report presenting 43 cases of Hirschsprung disease in which there were two six year old was mostly neonate and infants [13].

The male preponderance observed in this series confirms the observation on gender distribution of this condition [10]. The extent of the disease in this report showing ultra-short segment involvement of the rectosigmoid in the elderly patient possibly provided the explanation why the patient was able to cope with chronic constipation for the length of time. The hypertrophied active normally innervated proximal colon was perhaps able to overcome the prolonged partial colonic obstruction in this patient. There was no case of long segment or total colonic aganglionosis in this series, such cases would have probably presented much earlier.

The definitive surgical procedure in three patients (75%) of the cases was low anterior resection while one patient (25%) had Swenson Abdomino-perineal pull-through procedure, in which the distal aganglionic bowel was resected with preservation of continence by protecting the anorectal sphincters. Various other procedures such as Duhamel procedure [15], Soave procedure [16] and Lynn procedure [17] which have been used successfully in the surgical management of the childhood Hirschsprung disease have met with varying degrees of complications and treatment failure [2]. Low anterior resection (State procedure) had in the last 50 years [2] been employed for adult Hirschsprung disease and review of literature showed an average of 72% success rate and 6% mortality with this procedure [18]. Long term result outcome for surgical treatment of adult Hirschsprung disease is classified as good if there is

complete faecal continence with rare use of laxative and poor when there was continued reliance on mechanical dis-impaction with no improvement over the preoperative condition [2]. In our series out of the three patients who had anterior resection as a definitive procedure there was a post operative case fatality in the oldest patient. This could be attributed to pre-existing renal and cardiac co-morbidity. The other two patients who had low anterior resection (State procedure) were followed up for 4 years or more and were adjudged as having long term good result. Recent reports have advocated anorectal myectomy preceding low anterior resection in the treatment of adult Hirschsprung's [19,20]. This is thought to cause low morbidity and technical ease of dissection because of avoidance of deep pelvic dissection, it also accomplishes the removal of the most distal rectal aganglionic segment. The anorectal myectomy was not added to the procedure performed in these three patients however the result were still very good.

The last patient in our series had Swenson procedure. He had no post operative complications and although still early, a good outcome result after having being followed up for six months. Swenson procedure has been associated with excellent results in the hands of a few surgeons which has been difficult to reproduce both in children and adults. In 1975, Swenson *et al.* [21] reported their collective results with 282 patients including 71 adults and noted 5% incidence of anastomotic leak and 90% excellent long term results. No patient became impotent. These results have not been duplicated in adults by other surgeons [2].

Clearly, there is no obvious best choice of a surgical procedure for the treatment of Hirschsprung's disease in the adolescent and adult. The endorectal pull through procedure, the Duhamel-Martin procedure, and anorectal myectomy followed by low anterior resection are all associated with very good long term results. A successful outcome depends very much on the surgeon's experience with the procedure chosen.

It is also noted that the outcome is far better in adolescents and young adults as opposed to the elderly patient presenting with Hirschsprung disease as seen in this series, we also advocate that a two stage procedure with initial decompressive colostomy before definitive procedure has the tendency of improving postoperative morbidity and mortality in adult Hirschsprung's disease

Conclusion

Late presentation is a common feature of disease presentation in low resources countries like Nigeria, this worsens the outcome of surgical treatment with increased morbidity and mortality. Adult patients with long standing refractory constipation and recurrent use of laxative, enema saponis or manual disimpaction to relieve constipation should be suspected of having adult Hirschsprung's disease and should be investigated appropriately.

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