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A RARE PRESENTATION OF TWO COLORECTAL ANOMALIES: ANORECTALMALFORMATION CO-EXISTING WITH HIRSCHSPRUNG'S DISEASE IN IBADAN

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ABSTRACT

Background: **The association of Hirschsprung's disease with anorectal malformation (ARM) is both diagnostically and surgically challenging, in addition to it being a rarity. Symptoms attributable to post-repair of ARM may mask the underlying Hirschsprung's disease (HD) and become worse after closure of stoma. We aim to highlight this rare finding, the challenges associated with the management and the consequent multiple surgeries the patient underwent.**

Case presentation: **We report the case of a seven-year-old boy who had a diverting Devine descending colostomy for high ARM and was followed up until he had a posterior sagittal anorectoplasty (PSARP). He thereafter re-presented with complaints of intermittent constipation which was not responsive to rectal washout necessitating a levelling colostomy and subsequently an abdominal Swenson's pull-through procedure. Postoperative period was complicated with a colo-urethral fistula (Urethroscopy findings https://youtu.be/lxzyp1uHFpE?feature=shared) necessitating multiple staged surgeries.**

He has been followed up in clinic for over one-year post-surgery with complaints relating to frequent bowel motion, passes well-formed stool about three times daily.

We report a rare case of HD associated with ARM, highlighting the delay in diagnosis, the associated morbidities requiring multiple surgeries and the challenges encountered in the management of the patient.

Conclusion: **Hirschsprung's disease in a patient with anorectal malformation is a very rare occurrence, which can be fraught with delayed diagnosis and consequences as a result of the previous anorectoplasty. There is need to have a high index of suspicion and we hope this report will help raise the awareness of this association.**

Keywords: Anorectal malformation, Coexisting anomalies, Hirschsprung's, Ibadan, Nigerian.

INTRODUCTION

The association of Hirschsprung's disease (HD) and anorectal malformation (ARM) is rare. The exact incidence of this association is not known but HD coexisting with ARM has been reported in 2.3 to 5.6 % of patients with ARM.[1-3]

This association poses both a diagnostic as well as a surgical challenge as depicted in this index presentation. Symptoms such as constipation, straining, faecal soilage from stricture or anal stenosis attributable to the repair of the ARM may mask the underlying HD in both low and high ARM. The stoma for high ARM is usually fashioned proximally in the descending colon/sigmoid colon that is likely to be "ganglionated" which implies that the aganglionic segment distal to the stoma is reconnected and hence problematic after closure of stoma[4].

We aim to highlight a rare association of two major congenital colorectal malformations and illustrate the challenges encountered in the management of the patient.

Case Presentation

The patient, a seven-year-old male, who was delivered to a 29-year old healthy woman. He developed progressive abdominal distension with failure to pass meconium and was observed to have an absent anus on the second day of life. This necessitated a diverting Devine descending colostomy and he was subsequently followed up until he had a posterior sagittal ano-

rectoplasty (PSARP) at the age of 19 months, and closure of colostomy at 22 months.

Three months post colostomy closure, he re-presented with complaints of intermittent constipation which was initially responsive to digital anal stimulation with a diagnosis of anal stenosis complicating PSARP, and anal dilatation with Hegar's dilators was then judiciously recommended as per protocol.

Five years post PSARP he presented again to the surgery outpatient clinic with progressive abdominal distension. Bimanual rectal examination showed an indentable faecaloma in the lower abdomen. Twice daily rectal washout was instituted and he was lost to follow up until twenty-one months later, when he re-presented with worsening of symptoms necessitating an admission.

Examination at that time showed a school age boy who was pale, stunted with a distended abdomen, and had an indentable oval mass in the lower abdomen (Figure 1). Rectal examination revealed an empty rectum.

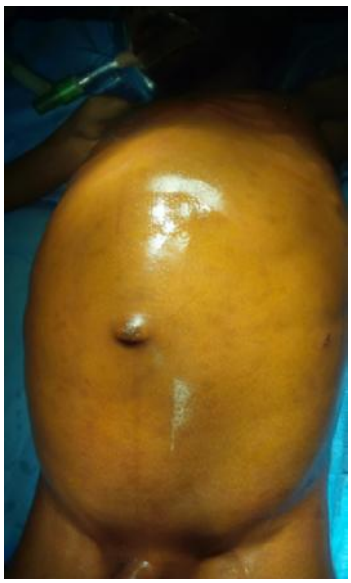

Figure 1. Clinical photograph of the abdomen at presentation at age six years.

A suspicion of Hirschsprung's disease coexisting with high-type anorectal malformation status post PSARP was entertained. Hypothyroidism and abdominal tuberculosis with background acyanotic congenital heart disease were also considered as differential diagnoses. The suspicion of tuberculosis was ruled out after full screening however, biochemical analysis showed features of sub clinical hypothyroidism and he was placed on L-thyroxine 75 mcg daily.

Chest radiograph done showed normal lung fields, however the heart appeared globular in configuration with appearances in keeping with a congenital heart disease with early decompensation (Figure 2).

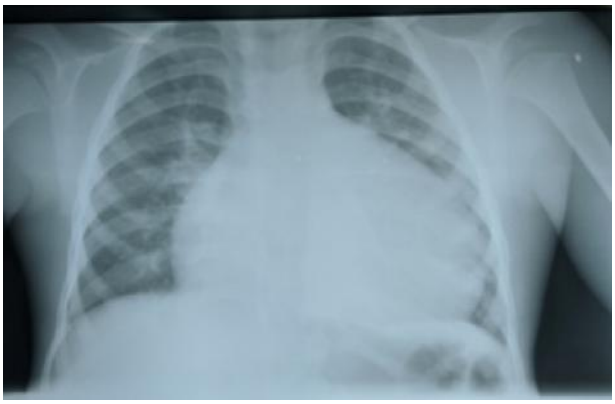

Figure 2. Chest Radiograph: Showing Cardiomegaly

Echocardiography showed features of asymptomatic pink tetralogy of Fallot, pericardial effusion of 35 millimeters on the right heart border and 18 millimeters on the left heart border.

Tube pericardiostomy was passed to drain the effusion and removed after 10 days post insertion. Pericardial fluid sent for gene-Xpert was negative for tuberculosis. He was then worked up for levelling colostomy.

Intraoperatively, he had resection of the dilated bowel forming a concentric mass (13 x 11 x 11 centimeters) filled with faeces (Figure 3a), with a Devine descending colostomy and triple biopsy done. Histology of the biopsy specimen were in keeping with a diagnosis of HD– colonic aganglionosis (Figure 3b, 3c).

The diagnosis of Hirschsprung's disease coexisting with anorectal malformation in a school child was now histologically confirmed.

He made a good recovery and was discharged home on the 5th post-operative day.

The patient was followed up in clinic and he was readmitted for definitive pull through surgery seven weeks post colostomy. He had an abdominal Swenson's pull-through with a colo-anal anastomosis, adhesiolysis with iatrogenic ileal perforations necessitating repair with end to end ileo-ileal anastomosis. He was discharged home on the 11th day post-surgery and followed up at the outpatient clinic where, he continued alternate day dressing of the abdominal wound with 10% povidone iodine on account of superficial surgical site infection. Healing by second intention occurred after six weeks post-surgery.

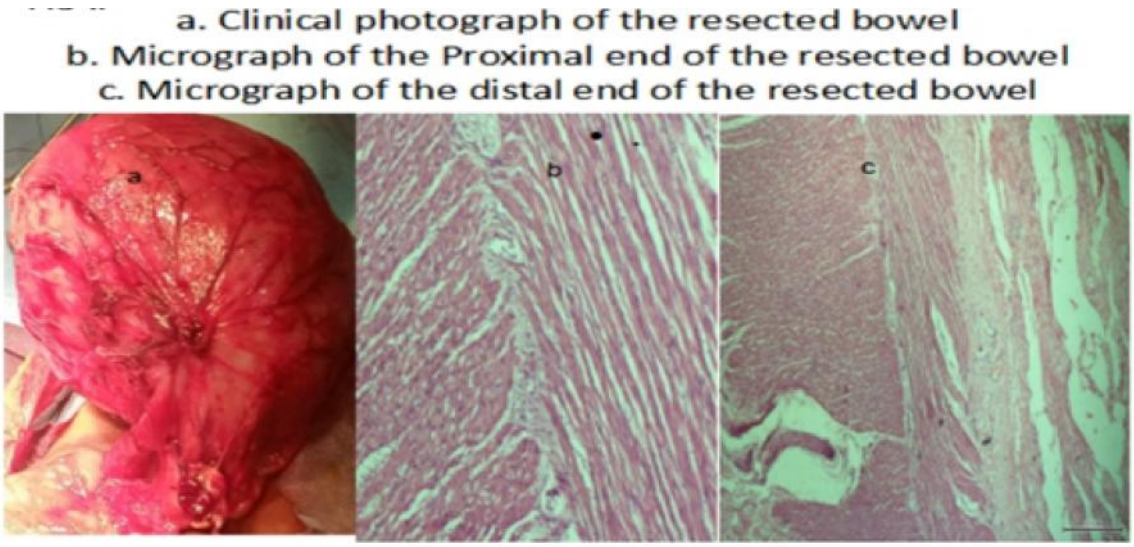

Figure 3. Intra operative photograph of the faecaloma (3a) with micrographs of confirmatory histology result of the biopsy specimens (3a and 3b) - showing presence of ganglion cells within the myenteric and submucosal plexuses as in figure 3b and absence of ganglion cells within the plexuses with hypertrophied nerve trunks as in figure 3c.

The result of the pull through specimen retrieved was in keeping with the rectal biopsy finding of HD, as shown in the micrographs below (Figure 3b, 3c).

During the follow up, he developed leakage of urine per rectum and faecouria with a suspicion of a colo-urethral fistula status post abdominal Swenson's pull through. (Figure 4). Seven months after the Swenson's pull through, he was worked up for a redo surgery and had combined abdominoperineal pull through with resection of a thick-walled inter-loop abscess and stenotic segment of the previously repaired ileo-ileal anastomotic site following an iatrogenic injury and a neo end-to-end ileo-ileal anastomosis was established with a tube caecostomy to protect the neo colo-anal anastomosis and a suprapubic cystostomy done. He developed low output enterocutaneous fistula and surgical site infection while on admission and was well managed with routine wound dressing and was later discharged home three weeks after surgery.

Two months post-surgery, his care givers noticed leakage of urine from the rectum and a diagnosis of a recurrent colo-urethral fistula post-surgery was made.

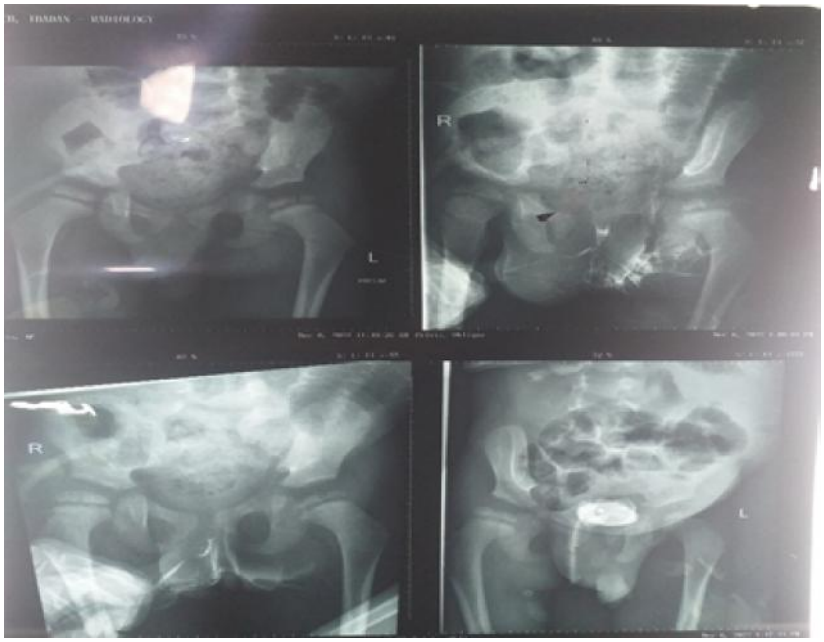

Figure 4. MCUG and Retrograde urethrogram showing a Recto urethral fistula.

He was readmitted seven months after the redo surgery for urethroscopy (Figure 5): Urethroscopy images showing a recto membranous urethra fistula. https://youtu.be/lxzyp1uHFpE?feature=shared" https://youtu.be/lxzyp1uHFpE?feature=shared) plus perineal exploration and repair of rectourethral fistula. Findings were a normal anterior urethra, with a fistulous connection between the membranous urethra and the anterior wall of the distal third of the neorectum. He subsequently had urethroplasty with good outcome and was discharged home nine days post operation and has since then been followed up in clinic.

The patient has been followed up in clinic for over one-year post-surgery with complaints limited to frequent stooling: he moves his bowel about three times a day of well-formed stool.

DISCUSSION

Simultaneous occurrence of both Anorectal malformation (ARM) and Hirschsprung's disease (HD) are rare.[4] A few cases have been reported in Europe and Asia, and in Zaria, Northern Nigeria. In a systemic review in 2013, of 38 articles (90 cases), the reported incidence was 2%.[4] Similarly, gender was reported in 63 cases, with 30 males (48%) and 33 females (52%), [4] which shows no gender predilection in patients with ARM coexisting with HD even though the index patient is a male. Nearly 35% of the initial colostomies performed in patients with ARM with HD are created in the aganglionic segment and in nearly 60% of the cases, the diagnosis of HD is not known at the time of operative ARM correction, leading to a median delay of eight months for the diagnosis of HD from the initial diagnosis of ARM and also a median delay of 17.5 months in the operative correction of HD in these patients. [4] Therefore, anything that points toward a possible association of HD in cases of ARM can help reduce the morbidity, delay in diagnosis as well as the surgeon's difficulty. In the index patient, a delay of six years eight months was noted for the diagnosis of HD from the initial diagnosis of ARM and a delay of about two months in the operative correction of HD was noted as well. The delay in diagnosis in this patient can be attributed to the initial response obtained from non-operative measures for colonic decompression (digital anal stimulation and rectal washout). However, because of the associated morbidities, it took over one year four months to have all the corrective surgeries for associated complications completed in this patient. In a series reported by Lister[5], he described firm, thick, blood vessels running in the mesentery of the colon of patients with HD, mostly conspicuous in the junctional zone. Another series reported abnormal arteries in 20 of 62 cases with HD at microscopic examination that were mostly located in the histological transition zone and were cited in the submucosa in 100% of cases.[6] The incidence of these abnormal arteries were observed to be higher the older the patients were at the time of resection (75% in >3 years of age).[7] The authors hypothesized that this abnormal pattern could expand proximally up to the mesenteric vessels as observed by Lister and that this histological finding was consistent with 'adventitial fibromuscular dysplasia' described by Stanley *et al.*[5,6,8] Grossly abnormal tortuous vessels have also been reported in the segmental dilatation of the intestine.[9.10] Like Lister, we also observed tortuosity of the vessels in the transition zone ramifying round the concentric mass in the index case during the time for levelling colostomy (Figure 3a), the presence of these can be further explained by the delay in diagnosis of HD at over 6 years due to the ARM.

The biopsy specimen from this site was aganglionic which is similar to findings by Lister in his series. Furthermore, we observed that the tortuosity in these vessels persisted till the time of pull-through in the patient even after the creation of a de-functioning stoma. This implies that the tortuosity was not secondary to the congenital bowel obstruction, but rather an inherent developmental abnormality of these vessels especially in short segment HD or when the transition zone extends beyond the sigmoid colon. In the review by Hofmann and Puri,[4] the extent of aganglionosis was reported in 49 cases and included classical rectosigmoid disease in 36, long segment aganglionosis in five, total colonic aganglionosis in seven and total intestinal aganglionosis in one patient. The index patient had a classical rectosigmoid disease.

Some patients with ultrashort segment disease are likely to be treated simultaneously during the PSARP. In over 60 % of cases, the diagnosis of an associated HD is not known at the time of ARM repair, which is like our experience; only 10 cases had been reported in literature on simultaneous pull-through for both ARM and HD.[2,11-13]

We believe that cases of high ARM associated with HD are more complex to treat. These patients have an absent anus, an aganglionic rectosigmoid segment of varying length, and a malformed sphincter mechanism. Thus, to improve continence, preservation of the distal bowel (rectum) as a reservoir is essential. A subsequent retrorectal (Duhamel) procedure which involves minimal burden for an already compromised anal sphincter and bladder innervations has been recommended. However, our patient had a Swenson's pull through.[14,15] The patients may also benefit from slowing down the colon.

Up to 25% of cases of ARM with HD reportedly have a syndromic association, namely: Currarino syndrome, Down's syndrome, Cat-eye syndrome, and Pallister-Hall syndrome.[2,4,16] In addition, HD is reported in 17% cases of ARM with coloboma of the iris.[17] There was no apparent syndromic association in the index case.

The diagnosis of HD is established in 15% of patients within the first month of life, in 40-50 % in the first three months, in 60% at one year of age and 85 % by the age of four years.[18] However, in the setting of an associated ARM, the diagnostic dilemma changes the tide and depicts the index patient whose diagnosis was made after the age of six. Therefore, a high index of suspicion is very important, and each patient treated carefully and wholesomely is key to early diagnosis.

CONCLUSION

Hirschsprung's disease in a patient with anorectal malformation is a very rare occurrence, which can be fraught with delayed diagnosis and consequences as a result of the previous surgical treatment offered for the anorectal malformation, vis-à-vis adhesions involving the large and small bowels and adjacent organs including the urinary tract. There is need to have a high index of suspicion and we hope this report will help raise the awareness of this association in our sub region as we recommend careful histological examination of the resected distal end of the rectum or colon for aganglionosis following a PSARP procedure.

REFERENCES

1. **Watanatittan S**, Suwatanaviroj A, Limprutithum T, Rattanasuwan T. Association of Hirschsprung's disease and anorectal malformation. J Pediatr Surg 1991; 26:192–195.
2. **Eltayeb AA**, Refaiy A. Association of Hirschsprung's disease with anorectal malformations: the early alarming signs for diagnosis and comorbidity related to this association. J Pediatr Surg 2014; pii: S0022-3468 (14)00279-6.
3. **Raboei EH**. Patients with anorectal malformation and Hirschsprung's disease. Eur J Pediatr Surg 2009; 19:325–327.
4. **Hofmann AD**, Puri P. Association of Hirschsprung's disease and anorectal malformation: a systematic review. Pediatr Surg Int 2013; 29:913–917.
5. **Lister J**. Abnormal arteries in Hirschsprung's disease. Arch Dis Child 1966; 41:143–154.
6. **Taguchi T**, Tanaka K, Ikeda K. Fibromuscular dysplasia of arteries in Hirschsprung's disease. Gastroenterology 1985; 88:1099–1103.
7. **Taguchi T**, Suita S, Hirata Y, Hirose R, Yamada T, Toyohara T. Abnormally shaped arteries in the intestine of children with Hirschsprung's disease: etiological considerations relating to ischemic theory. J Pediatr Gastroenterol Nutr. 1994; 18(2):200-204.
8. **Stanley JC**, Gewertz BL, Bove EL, *et al.* Arterial fibrodysplasia. Histopathologic character and current etiologic concepts. Arch Surg 1975; 110:561–566.
9. **Takehara H**, Ishibashi H. Congenital segmental dilatation of intestine. In: Puri P, editor. Newborn surgery, 2nd ed. London: Arnold (Hodder Headline Group); 2003. Pp. 553–556.
10. **Kothari P**, Gowrishankar, Rastogi A, *et al.* Congenital segmental dilatation of colon with colonic atresia. Indian J Gastroenterol 2005; 24:123–124.
11. **Mahboubi S**, Templeton JM Jr. Association of Hirschsprung's disease and imperforate anus in a patient with 'cat-eye' syndrome. A report of one case and review of the literature. Pediatr Radiol 1984; 14:441–442.
12. **Arbell D**, Gross E, Orkin B, Koplewitz BZ, Udassin R. Imperforate anus, malrotation, and Hirschsprung's disease: a rare and important association. J Pediatr Surg 2006; 41:1335–1337.
13. **Lukong CS**, Mshelbwala PM, Anumah MA, Ameh EA, Nmadu PT. Anorectal malformation coexisting with Hirschsprung's disease: a report of two patients. Afr J Paediatr Surg 2012; 9:166–168.
14. **Zundel S**, Obermayr F, Schaefer JF, Fuchs J. Hirschsprung's disease associated with total colonic agenesis and imperforate anus – case report and review of the literature. J Pediatr Surg 2010; 45:252–254.
15. **Pant N**, Khan TR, Malhotra S, Kumar P, *et al.* Diagnostic suggestion and surgical consideration for Hirschsprung's disease associated with high anorectal malformation. Annals of Pediatric Surgery 2018, 14:78–82
16. **Laje P**, Fenton SJ, Peranteau WH. Total colonic Hirschsprung's disease and anorectal malformation in a baby with Pallister-Hall syndrome. J Pediatr Surg Case Rep 2013; 1:308–310.
17. **Brisighelli G**, Bischoff A, Levitt M, *et al.* Coloboma and anorectal malformations: a rare association with important clinical implications. Pediatr Surg Int 2013; 29:905–912.
18. **Chirdan LB**, Uba AF (2006) Hirschsprung's disease presenting in the neonatal period in Jos, Nigeria. Nig. J. of Surgical Research 8: 62-64.