Case Report

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Anorectal malformation coexisting with Hirschsprung's disease: A report of two patients

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ABSTRACT

Anorectal Malformation (ARM) and Hirschsprung's Disease (HD) are common causes of congenital intestinal obstruction in children. Simultaneous occurrence of both conditions is rare. Few have been reported in Europe and Asia, but we have no knowledge so far of such report from Nigeria. We present two patients managed in our centre to highlight the challenges of management of this uncommon coexistence. The first patient was a 5-year-old girl who was referred to us with intestinal obstructive symptoms despite an apparently adequate sized ectopic anus. She had colostomy and rectal biopsy, which confirmed HD. She had corrective surgery performed through a posterior sagittal approach. She did well post operatively. The second patient was a 3-year-old girl who presented with features of intestinal obstruction, had laparatomy and was also referred to us. It was observed in the referral hospital during laparatomy, to have features of HD and rectal atresia intraoperatively. She had colostomy done and rectal biopsy performed at the same time, which confirmed the diagnosis of HD. She had simultaneous correction of both conditions through a posterior sagittal approach. She was in good condition at follow up. It was therefore recommended that a high index of suspicion of HD, should be entertained while managing patients with anorectal malformation.

Key words: Anorectal malformation, Hirschsprung's disease, posterior sagittal approach

INTRODUCTION

Anorectal malformation and Hirschsprung's disease are common causes of intestinal obstruction in children. Each of these conditions has a frequency of 1: 4000-5000 live births.[1,2] The frequency of both conditions coexisting has not been reported in the literature.

The association between anorectal malformation and Hirschsprung's disease was first noted and reported in literature by Kiesewetter *et al*. in 1964.[3] Since then there has been few reports from around the globe.[4-8] None of such reports is from Nigeria.

Both conditions coexisting could pose a diagnostic challenge when managing patients with intestinal obstruction. Intestinal obstruction from each of the conditions may result in morbidity and mortality during management.[8] The morbidity and mortality could even be more if both conditions coexist.

This article therefore presents management challenges faced in two patients with anorectal malformation coexisting with Hirschsprung's disease seen in a tertiary hospital in Northwestern Nigeria.

CASE REPORTS

Case 1

A 5-year-old girl presented to our hospital for definitve surgery, with Ectopic anus and a divided sigmoid colostomy. She was initially seen in a tertiary centre, where she had colostomy done due to inability to decompress via the ectopic anus. This was associated with recurrent episodes of constipation and abdominal distension. The perinatal period was essentially normal. The parents could not remember whether the child had delayed passage of meconium.

At presentation, she was well preserved, weighed 17 kg. Her cardiorespiratory status was normal. Abdominal examination revealed full abdomen with a divided sigmoid colostomy. The abdomen was not tender and no palpable organomegaly. Bowel sounds were normal.

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Perineal examination revealed an ectopic anus that admitted Hegar's dilator size 13. She had normal female external genitalia. Her vertebral column, pelvis and limbs were normal.

A suspicion of ectopic anus associated with Hirschsprung's disease was entertained.

She had rectal biopsy via the ectopic anus which confirmed aganglionosis.

She had corrective surgery via the posterior sagittal route. The intra-operative findings were; ectopic anus, dilated proximal rectum. She had distal bowel resected, colostomy take down and coloanal anastomosis done in one layer.

Post-operative recovery was good. She was discharged home in good condition with normal anorectal function. Histology of resected colonic segment (40 cm), showed aganglionosis of the distal colon, with ganglion cells in the proximal bowel (Faecal stoma or fistula).

At follow-up, the patient had remained normal with normal bowel function.

Case 2

A 3-year-old girl was referred to our Unit from the National Hospital at 14 months of age with a diagnosis of rectal atresia and Hirschsprung's disease.

She presented at the referral hospital at 2 days of life with inability to pass meconium since birth and also features of intestinal obstruction.

She had a laparotomy done during which a small colon distal to the splenic flexure was noted and biopsies taken from the collapsed portion and a colostomy fashioned. Histology of the biopsies revealed aganglionosis and was therefore referred to us, for expert management. Her perinatal history was essentially normal.

At presentation, she was not pale and weighed 6.0 kg. Her cardio-respiratory status was normal. The abdomen was full, moved with respiration with a loop transverse colostomy at the left hypochondrial area. The abdomen was not tender, and no palpable organomegaly. The bowel sounds were normal. She had normal female external genitalia. Anorectal examination revealed well located anus with rectum ending blindly 4 cm from the anal verge.

With diagnosis of rectal atresia and Hirschsprung's disease from the referral hospital, further evaluation was done in our centre. She had biopsies take from the distal and proximal stomata, which revealed aganglionosis and ganglion cells, respectively.

She had colostomy takedown and both conditions tackled via the posterior sagittal route. Intra-operatively the findings were; atretic rectum 6 cm distally, normal small bowel. She had colostomy takedown, with segmental resection of the distal stoma and colon including the atretic distal rectum (resected colon 40 cm). The proximal colon was then used for the coloanal anastomosis done in one layer.

Post–operative period was uneventful and she was discharged home in good condition.

At follow-up, patient was found to be normal with good bowel function. Histology of resected rectal segment confirmed aganglionosis, the proximal stoma used for coloanal anastomosis had normal ganglion cells.

DISCUSSION

The two patients presented in this series are both females and they will form a basis for discussion.

The simultaneous coexistence of anorectal malformation and Hirschsprung's disease is rare. The aetiology of both conditions remains obscured and unknown. There are postulates implicating genetic, familial and environmental factors. No single factor or groups of factors have been noted to be responsible for the simultaneous occurrence of anorectal malformation and Hirschsprung's disease. The vascular theory may be responsible for simultaneous occurrence of conditions. Puri *et al*. in search of for the aetiology of Hirschsprung's disease, concluded that knowledge about the aetiological basis of this disease was poorly understood.[9] Taquchi *et al*. in their study noted abnormally shaped arteries in the intestine of children with Hirschsprung's disease and based their aetiological consideration to ischaemic theory.[10]

Each of the conditions is responsible for morbidity and mortality in paediatric surgical practice. The coexistence of the two conditions may aggravate or increase the morbidity and mortality in children. The consequence of diagnosing one condition without the other when there is this simultaneous coexistence may be grievous in the management of these patients.

In the two patients presented, such simultaneous

occurrence was not considered ab initio. This is due to rarity of the condition making suspicion obscure in management.

Morbidity and mortality might have ensued, if this suspicion of coexistence of both conditions was not born in mind for our patients.

The two patients had both conditions tackle via the posterior sagittal approach. This idea was extrapolated from the knowledge of exposure given by incision of posterior sagittal anorectoplasty. This was found to be quite useful in our patients. The posterior sagittal incision can be used in the management of Hirschsprung's disease.[11]

In conclusion, the coexistence of anorectal malformation and Hirschsprung's disease is rare but does occur. Strong suspicion, prompt diagnosis and the posterior sagittal approach are key factors in the management of both conditions.

We therefore recommend that patients with anorectal malformation and perineal fistula who fail to decompress after adequate treatment should be evaluated for Hirschsprung's disease. Also during colostomy for anorectal malformation, if the distal segment shows features of Hirschsprung's disease biopsies should be taken.

We are of the opinion that all patients with anorectal malformation be screened for Hirschsprung's disease with a view to establishing the prevalence of patients with the coexistence of both conditions in the environment.

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