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Entero-umbilical fistula mimicking patent vitello-intestinal duct as a rare presentation of Hirschsprung's Disease

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Introduction

Hirschsprung's Disease (HD) is a developmental disorder of the intrinsic component of the enteric nervous system that is characterized by the absence of ganglion cells in the myenteric and submucosal plexuses of the distal intestine. Neonatal obstruction, delayed passing meconium and enterocolitis are not uncommon in the clinical presentation of affected neonates . However we experienced a rare presentation of HD which was mimicking a presentation of patent vitello-intestinal duct.

Case Description

A healthy 3 month old baby boy, never had history of delayed passing meconium, distension or enterocolitis previously. Unfortunately he presented with 2 weeks duration of omphalitis and faecal discharge from umbilicus in which further worsen with intestinal obstruction symptoms. His distended abdomen showing erythematous skin surrounding an umbilical polyp, discharging faecal material. A patent vitello-intestinal duct is suspected. Furthermore, gross bowel dilatation seen from plain abdominal radiograph, mandate for urgent laparotomy to release obstruction. During laparotomy, entero-umbilical fistula communicating the umbilicus with a very dilated sigmoid colon seen. The rectum appeared collapsed. The upper sigmoid brought out as stoma while the mucosal edge of stoma sent for histopathological examination (HPE) and it revealed as ganglionic. Suction rectal biopsy was done later and revealed aganglionic segment. Duhamel's procedure was done, sigmoid colostomy was brought down and anastomosed at anal canal. HPE of the resected rectosigmoid segment intraoperatively confirmed the absence of ganglionic cells at distal 17mm which further supported the diagnosis.

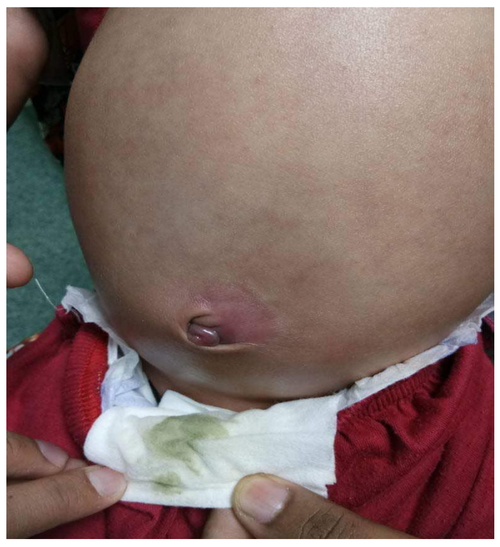

Fig 1. Umbilical polyp discharging greenish faecal materials. Noted that the abdomen is very distended.

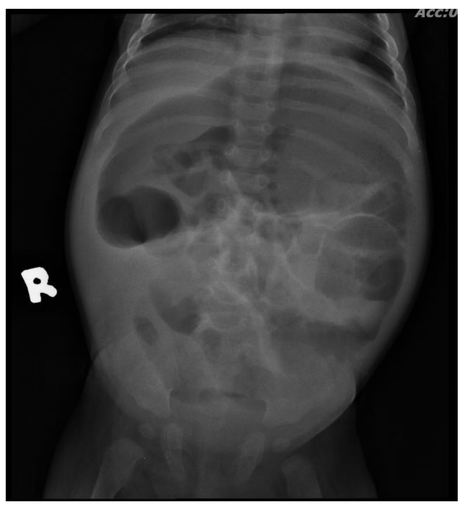

Fig 2. Dilated bowel seen on plain abdominal radiograph

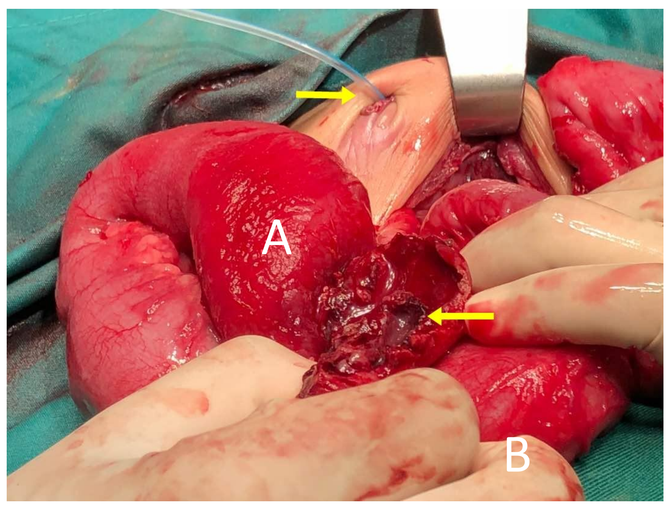

Fig 3a. Entry site of feeding catheter used as probe through the umbilicus
Fig 3b. Exit site of probe into the lumen of dilated sigmoid colon

Discussion

A study by *Bradnock et al* described abdominal distension and bilious vomiting were the most common clinical features at presentation [1], besides that, there are also other presentation has been reported. The 'classic triad' of billious vomiting, abdominal distension and delayed passage of meconium was evident only in 26% of cases [1]. *Sarioglu et al* reported 2 cases of colonic volvulus, a caecal volvulus in which occurred in 5 day old neonates and the other one sigmoid volvulus on older 11 year old boy [2]. They conclude the sigmoid volvulus have short segment of HD. If the mesosigmoid is freely mobile, the dilated ganglionic sigmoid colon seems to have a predilection to sigmoid volvulus. However, if the involved segment extend beyond sigmoid colon, other segments such as transverse colon and caecum may undergo volvulus in the presence of anatomic factors [2]. A systematic review state that congenital megacolon is the most common cause of intestinal obstruction in neonates, with the vast majority of cases of HD being diagnosed in this period [3]. In a few cases, however, the condition may be masked when constipation is successfully managed by laxatives and enemas, allowing some patients to reach adulthood before the diagnosis is made [3]. Furthermore, failure of internal sphincter relaxation is the cause of manometry failure when the biopsy and fluoroscopy study excluded, used as diagnostic tool in cases of 'controversial' ultrashort HD where those patient presented with chronic constipation [4]. Appendiceal perforation also describe as rare modes of HD with the mortality associated seems to be higher [5]. It should be differentiated from a perforated appendicitis, as inflammation begins from mucosa towards serosa and perforation usually occurs at tip, however in HD the mucosal infiltration is minimal, and periappendicitis is more severe. In addition, *Roshkow et al* also describe the neurocristopathy manifested by Ondine's curse (congenital hypoventilation syndrome) and congenital neuroblastoma with HD [6].

Conclusion

This report identified variable presentation of HD. In summary, we wish to report a rare presentation, where our patient presented with entero-umbilical fistula mimicking patent vitello-intestinal duct, in hoping to provide aid and better understanding for future research.

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