Case Report

Atresia of the Colon Associated with Hirschsprung’s Disease

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Abstract

Atresia of the colon is a rare anomaly with an incidence of between 1:20,000 and 1:66,000 live births being reported. Hirschsprung’s disease association with Colonic atresia is usually diagnosed after several failures of intestinal anastomoses. We herein report one of the first patients in the literature diagnosed before a therapeutic challenge. A 2-day-old female was admitted with severe abdominal distention, bilious vomiting and failure to pass meconium. A distended abdomen accompanied by hypoactive bowel sounds was also observed. Abdominal X-ray revealed increased intestinal gas, mainly in the colon. Type III, atresia of the colon at the level of the splenic flexure was found at laparotomy. A temporary double-barrel colostomy was completed, and she was discharged from hospital on the tenth day after operation without any complications. At the age of 3 months, due to the aspect of the distal colon, a rectal biopsy was performed and aganglionosis was confirmed. The combination of intestinal aganglionosis and colonic atresia is extremely rare. The concomitance of colonic atresia and aganglionosis is calculated to be in 1 in 10 million live births. Wilson, et al. claims that 80 percent of infants with colonic atresia have associated gastrointestinal anomalies. These defects include rotation and fixation anomalies. However, aganglionosis and intestinal neuronal dysplasia should be taken into account as well. When both diseases are combined, the etiology is still uncertain and several etiologies have been suggested. The association should be suspected in all cases of colonic atresia and rectal biopsies are advocated at the primary operation in patients with atresia of the colon.

Keywords: Hirschprung disease, intestinal atresia, neuronal intestinal dysplasia


Introduction

Atresia of the colon is a rare anomaly with an incidence of between 1:20,000 and 1:66,000 live births being reported.1,2 Only 20 patients were previously communicated combined with the Hirschprung’s disease.3,4 Akgür, et al. reported in 1993 the first patient with associated Hirschprung’s disease in the literature diagnosed before attempting intestinal anastomosis for stoma closure.4 Before that era, these patients underwent failed reconstructions until it became obvious that colonic atresia should be generally screened for Hirschprung’s disease with a rectal biopsy.3 This report presents a new case and review the literature, both of which raise interesting points regarding the management of this rare disease.

Case Report

A 2-day-old full-term female baby with a birth weight of 3200 g was born to a 28-year-old mother. She was admitted with severe abdominal distention, bilious vomiting and failure to pass meconium. It is important to note that she was discharged from the center without having exact information about bowel depositions. The family history, maternal history and prenatal examinations were all unremarkable. No drugs or infectious exposure was documented during pregnancy. Physical examination revealed dehydration and respiratory distress with subcostal retraction. A distended abdomen accompanied by hypoactive bowel sounds was also observed. The X-ray examination of the chest and abdomen revealed increased intestinal gas, mainly in the colon (Figure 1). We did not have the advantage of a contrast enema in order to refine the diagnosis, as the radiology department is not equipped for diagnosis in newly born patients. Therefore, we had to act according to the intuition and the patient was operated on day 3. Type III, atresia (Grosfeld’s classification) of the colon at the level of the splenic flexure was found at laparotomy (Figure 2). The discrepancy in diameters between the proximal and distal segments was 10:1. The distal colon was foreshortened and coiled in the pelvis. A temporary double-barrel colostomy was completed, and then she was discharged from the hospital on the 10th day after operation without any complications. The biopsy of the proximal colon showed the presence of ganglion cells. At the age of 3 months, a rectal biopsy was performed because of the aspect of the distal colon and aganglionosis was confirmed. We have obtained the consent from parents to report the case, but after that, the child was not brought for further follow-up.

Discussion

The combination of intestinal aganglionosis and colonic atresia is extremely rare. Its simultaneous incidence is calculated to be in 1 in 10 million live births.1 Until 1993,2 the association was not recognized in the neonatal period, and the postoperative courses of colocolic anastomosis were complicated and troublesome. In the past, the association was only recognized when a primary anastomosis became functionally obstructed.4 Some researchers suggested that the association of Hirschsprung’s disease is extremely rare and rectal biopsy is not necessary before primary anastomosis. However, high clinical suspicion should be taken in order to rule out Hirschprung’s disease in children who present with colonic
Some authors claim that 80 percent of infants with colonic atresia have another gastrointestinal anomaly, but aganglionosis and intestinal neuronal dysplasia should be taken into account as well. If the combination of colonic atresia-Hirschsprung’s disease is not diagnosed early, it conduces to elevated morbidity and mortality, especially 72 hours after delivery. We want to stress the importance of diagnosing both conditions just to avoid annoying complications like anastomotic dysfunction or leakage. The safest way to proceed is to perform a proximal decompressing stoma.

The ultimate etiology of neonatal intestinal atresia is well defined and the theory of an antenatal vascular insult to the intestine is accepted by the majority of the authors. However, when colonic atresia and Hirschsprung’s disease are combined, the etiology is still uncertain and several etiologies have been suggested: from the random theory to vascular alterations.

Obviously, colonic atresia should lead to urgent surgery because of the closed loop obstruction, which leads to bowel necrosis and perforation. The preoperative contrast enema should have been performed to diagnose colonic atresia and visualize the level of the distal segment. However, our patient only received a plain babygram followed by the decision that she needed surgery. Several cases have been previously reported of colonic atresia associated with gastrochisis, facial asymmetry and/or ophthalmologic problems, but the association with Hirschsprung’s disease is very unusual.

In summary, we reported a rare case of Hirschsprung’s disease associated with colonic atresia. It is suggested that there may be a connection between these two conditions. We also stress that, although very rare, this malformation should be considered when dealing with neonatal bowel obstruction. The association should be suspected in all cases of colonic atresia and rectal biopsies are advocated at the primary operation in patients with atresia of the colon.

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