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 Hirschsprung’s disease.3,4 Akgitir, et al. reported in 1993 the first patient with associated Hirschsprung’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 Hirschsprung’s disease with a rectal biopsy.5 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. 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 aganglioneosis was confirmed. The combination of intestinal aganglioneosis and colonic atresia is extremely rare.6 The concomitance of colonic atresia and aganglioneosis 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, aganglioneosis 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.

Discussion

The combination of intestinal aganglioneosis and colonic atresia is extremely rare. Its simultaneous incidence is calculated to be in 1 in 10 million live births.1 Until 1993,4 the association was not recognized in the neonatal period, and the postoperative courses of colocolonic anastomosis were complicated and troublesome. In the past, the association was only recognized when a primary anastomosis became functionally obstructed.5 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...
atresia. 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.2,3 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.7 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.5 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.2,3,7

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