Ileal Atresia and Total Colonic Aganglionosis

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Abstract

Herein, the authors report a case of ileal atresia who failed to have good bowel function after primary anastomosis. After the leakage of the revised anastomosis, a thorough pathological review found distal aganglionosis. An ileostomy followed by an ileocolic patch operation were performed for temporary decompression, awaiting intestinal adaptation. A definitive pull-through was performed, eight months later. The child also had bilateral cleft lip together with complete cleft palate, and hemivertebrae of the thoracic spines. A cluster of malformations is unusual in a single patient.

Key word : Ileal Atresia, Total Colonic Aganglionosis, Multiple Congenital Anomalies

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Although extremely rare, associated Hirschsprung's disease is occasionally found in neonates with intestinal atresia. Such an occurrence has been reported and explained as an embryonic sequence, during the migration of the ganglion cells. However, presence of the two conditions together with vertebral defects and cleft lip/cleft palate is unusual. A single case is reported with all these anomalies and the pathogenesis and surgical implications are discussed.

CASE REPORT

A three-day old male infant was referred to us from a provincial hospital with a preliminary diagnosis of intestinal obstruction. The child was born

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vaginally with a birth weight of 3,050 grams and good APGAR score. Bilateral complete cleft lip and cleft palate were the only external anomalies detected. The patient developed abdominal distension and bilious vomiting on the first day. Abdominal X-rays revealed multiple bowel-loops dilatation, different height of air-fluid level and absence of air in the distal bowel, suggesting a complete low-gut obstruction. The hemivertebrae was found at the 7^{th} - 8^{th} thoracic spines, resulting in crowding of the adjoining ribs. Bilateral absence of the 12^{th} rib was also noted (Fig. 1).

Laparotomy disclosed an ileal atresia with a large mesenteric gap. A single small artery on the mesentery fed the blind-ended proximal pouch, which looked possibly gangrenous (Fig. 2). The proximal bowel length before resection was 103 cms. The ileum distal to the obstructing site looked small, but well patent. The entire large intestine was found to be small and poorly fixed to the posterior abdomen. A tapering ileo-ileostomy was performed after a resection of the proximal ileum of approximately 25 cms.

The infant began to have bowel movement on the sixth post-operative day. The stool frequency ranged from 0-3 times a day, but at least once most days. However, enteral feeding could not be introduced because the patient continued to have a large gastric content. A barium enema was done, showing a small colon and narrowed anastomosis (Fig. 3). A re-exploration and anastomotic revision was scheduled on the 53^{rd} post-operative day. Five days later, the second anastomosis had leakage, necessitating a repeated laparotomy for a Bishop-Koop ileocolostomy. The histopathology review of the distal

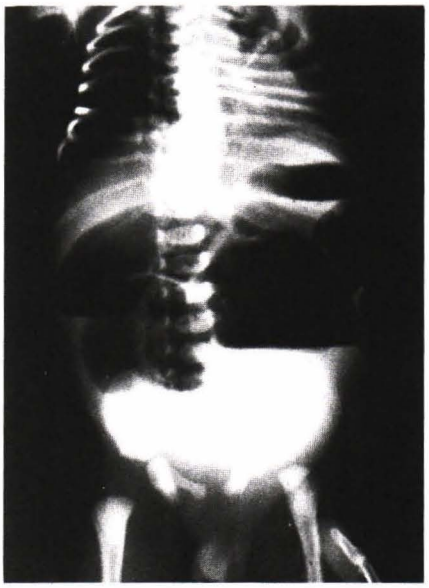

Fig. 1. Plain abdominal X-rays on the second day after birth, demonstrating intestinal dilatation, air-fluid level in different heights, hemivertebrae and abnormal ribs.

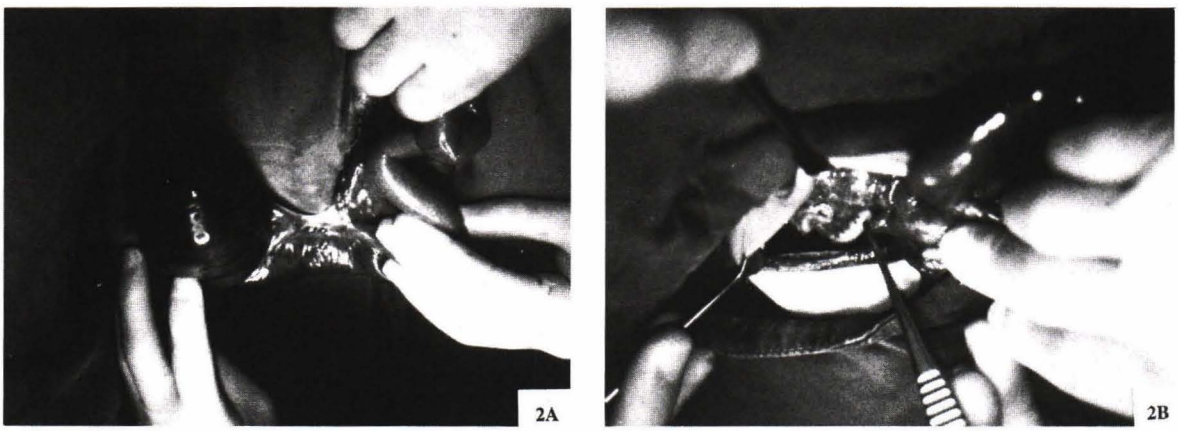

Fig. 2. Gross finding of the ileum A) the proximal bowel supplied by only one vessels, B) microcolon at the distal bowel, which went straight down to the pelvis without proper fixation.

Fig. 3. Barium enema on the sixth post-operative week, showing the narrowed anastomosis and the persistent microcolon. Note the alignment of the colon that presented no anatomical curvature.

ileum, cecum and the appendix demonstrated aganglionosis. An ileostomy with distal mucous fistula was then established, later.

Unfortunately, the child exhibited short bowel syndrome. The ileostomy content was approximately 200 ml/day. A Kimura's colonic patch was considered two months later. Enteral feeding was then possible, although the parenteral nutrition could not be totally weaned off. A modified-Duhamel's pull-through operation was done at nine-months and the small bowel adaptation reached acceptable enteral autonomy. The parenteral nutrition was discontinued two weeks after the pull-through operation. The child was discharged on his 309th admission day. His body weight on the discharge day was 7.8 kg.

Total repair of the cleft lip and cleft palate was done one month before the pull-through operation. The chromosome study was normal. Ultrasound showed no abnormality in the urinary system. The histology of all surgical specimens confirmed the diagnosis of total colonic aganglionosis. The child is now in a long-term follow-up program for multiple congenital anomalies. He is doing well and has good weight gain. Despite the vertebral anomaly, no neurological defects were experienced.

DISCUSSION

Associated congenital anomalies are found in 11.3 to 60 per cent of infants born with jejunoileal atresia[1-3]. The majority are confined to the gastrointestinal system, such as abnormal rotation and synchronous atresia. Mechanical obstruction and a defect in the innervation of the intestine rarely occur together. To the best of our knowledge, not more than twenty cases of jejunoileal atresia and Hirschsprung's disease have been reported in the English literature, and none of those cases had associated vertebral anomaly and cleft lip/cleft palate in the same patient[4].

The pathogenesis of these synchronous gastrointestinal anomalies has been explained by different theories[4-8]. All involve the embryologic sequences during the craniocaudal migration of the enteric neurons, which takes place between the 5th - 12th week following conception. Complete disruption of the intestine with mesenteric defect as in the presented patient, if it occurs before the arrival of the neural crest cells, may directly explain the distal aganglionosis. However, not every case with aganglionosis has this gap. Vascular accident is a widely accepted theory for the embryogenesis of intestinal atresia[9]. Such an event can be a common factor for the arrest of neural crest migration at the same distance as the atresia occurs. The presence of a short distance of normal innervation, distal to the atretic site, in some cases is opposed to this explanation[4].

In utero volvulus and intussusception were reported to be associated with aganglionosis[9,10]. The volvulus precedes the atresia and aganglionosis. The evidence of incomplete fixation and congenitally short bowel in our case agreed with this relation. On the other hand, the obstructing intestine secondary to aganglionosis may predispose the fixating bowel to twist[4]. This is supported by 12-25 per cent presence of cystic fibrosis in Caucasian jejunoileal atresia. The presence of a poorly fixed colon in a case of intestinal atresia can be a clue to aganglionosis[11].

Additional extra-intestinal anomalies are evidence that the patient has been subjected to an intrauterine insult that interfered with the formation of multiple organs[9]. There have been reports of

colonic atresia, aganglionosis and absent hand, and of the first two and omphalocele[8,12]. Hemivertebrae was found in 1/35 cases of jejunoileal atresia in one report, as was a cleft lip/palate[1]. However, the co-existence of those anomalies with aganglionosis in a single case has never been reported.

The clinical implication of the aganglionosis is that it causes anastomotic dysfunction[5,7]. The condition is not easily recognized because oral feeding in a case with jejunoileal atresia is naturally delayed. Careful inspection of the ganglion cells in the distal bowel should be the key to the diagnosis. Unfortunately, at the first operation, the authors did not send a specimen from the distal ileum for an examination. And because there were reports of ganglion cells distal to the atresia, it was suggested that a rectal biopsy should be done in case of failure of the bowel to function properly after corrective surgery[4]. Examination of ganglion cells in the vermiform appendix can also be an alternative[13].

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ลำไส้เล็กส่วนอิเลียมตีบและภาวะขาดเซลล์ปมประสาทอย่างสมบูรณ์

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ได้รายงานผู้ป่วยทารกแรกเกิดซึ่งมีภาวะทางเดินอาหารอุดกั้นจากการตีบของอิเลียม ทารกรายดังกล่าวรับอาหารไม่ได้และมีท้องอืดภายหลังการผ่าตัดเพื่อต่อเชื่อมทางเดินอาหารครั้งแรกเป็นเวลาหลายสัปดาห์ การผ่าตัดต่อเชื่อมทางเดินอาหารครั้งที่สองประสบปัญหารอยต่อรั่ว การทบทวนทางพยาธิวิทยาพบลำไส้ส่วนปลายขาดเซลล์ปมประสาท เข้าได้กับโรคเฮิร์ทซปรุง ซึ่งเป็นตลอดลำไส้ใหญ่ ทารกได้รับการยก ileostomy ไว้ชั่วคราวและทำผ่าตัดขั้นสำเร็จในเวลาแปดเดือนต่อมา นอกจากความพิการทั้งสอง ทารกรายนี้ยังมีปากแหว่งเพดานโหว่ และความผิดปกติของกระดูกสันหลังชนิด hemivertebrae การเกิดความพิการทั้งหมดนี้ในผู้ป่วยรายเดียวกัน ยังไม่เคยได้รับการรายงานมาก่อน

คำสำคัญ : อิเลียมตัน, โรคเฮิร์ทซปรุง, ปากแหว่งเพดานโหว่, ความพิการแต่กำเนิดหลายระบบ

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