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Case Report

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Ileal Atresia in Down Syndrome, A Rare Finding

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1. Introduction

A 37-week-old baby boy was born by cesarean section to a 40-year-old G7P6006 mother who tested positive for 21+ by noninvasive prenatal testing. At delivery, the baby had features of Down Syndrome, including low-set ears, bilateral epicanthal folds, a flat nasal bridge, a protruding tongue, a Simian crease on the right hand, an umbilical hernia, and excess skin on the nape of the neck. He presented with Apgar's of 7 and 8. He was admitted to the NICU requiring CPAP due to respiratory distress, kept NPO, and started on IV fluids. When feeds were started at the end of the first day of life, he was noted to have episodes of bilious emesis, with abdominal distention, and failure to pass meconium till 36 hours of life.

Therefore, a contrast barium enema was performed, which showed a rectum larger in caliber than the collapsed nondilated overlapping loops of colon which was only opacified to a lead point left mid abdomen. The contrast did not pass beyond this lead point. Because the barium did not move further than the lead point, meconium plug, ileal, or colonic atresia were considered in the differential diagnosis. He was transferred to a higher level NICU on day of life 3, namely Level IV, where he underwent an upper gastrointestinal study that demonstrated retention of contrast within microcolon and severely distended small bowel loops without contrast. He then was taken up for exploratory laparotomy, and ileal atresia was noted. It was resected and a small bowel-to-small bowel anastomosis was performed. The postoperative course was uneventful, he was started on feeds on day 5.

1.1. Operative Findings

The laparotomy identified a proximal ileal atresia at a distinct transition point, affecting approximately 10 cm of the ileum, with significant dilation. The ileal atresia was classified as a type 2 atresia by the Pediatric Surgeon which signifies intestinal separation but no mesenteric separation. The duodenum and jejunum appeared entirely normal, extending to the ligament of Treitz.

In the distal portion, a viable and patent segment of the distal ileum was observed, followed by a normal-looking cecum and appendix. The dilated 12 cm segment was excised and sent for pathological examination. No additional atresia's were found. Subsequently, a healthy ileo-intestine with a normal caliber and a small collapsed distal segment was achieved, concluding with an ileum-to-ileum anastomosis. He had short-term direct hyperbilirubinemia which improved subsequently, and the baby was discharged home on day 7 of life.

2. Discussion

Atresia, characterized by abnormal openings, constrictions, or passages within the body, can manifest throughout the bowel, presenting as either solitary or multiple lesions. In neonates, jejunoileal atresia stands out as a frequent finding second only to intestinal obstruction. The genesis of intestinal atresias often involves vascular accidents or the failure of bowel recanalization. Vascular lesions arise from disruption of mesenteric vessels, typically the superior mesenteric artery leading to ischemic necrosis of the fetal intestine, resulting in segmental resorption, i.e., a discontinuous bowel. However, local vascular compromise may also play a role in developing small bowel atresia. It is believed that conditions that may obstruct or constrict the blood supply to the intestine, including internal hernias, volvuli, and intussusceptions, may also trigger jejunoileal atresia.

Ileal atresia, a congenital anomaly, is associated with various risk factors that contribute to disruptions in fetal intestinal development. These risk factors encompass underlying intestinal pathology, thrombotic processes, and genetic predispositions. One risk factor is cystic fibrosis, which poses an elevated risk. Due to the formation of thickened meconium in cystic fibrosis, there is an increased chance of the potential development of a segmental volvulus, leading to jejunal or ileal atresia [1]. Cases of gastroschisis are often complicated by intestinal atresias, arising from either strangulation due to the fascial defect or volvulus of herniated bowel, leading to necrotic tissue absorption and subsequent atresia formation, commonly in the proximal jejunum [2].

Also, midgut volvulus, specifically Type IIIB atresia, can occur during gestation, causing the disappearance of a significant portion of the intestine nourished by the superior mesenteric artery, resulting in high jejunal or occasionally distal duodenal atresia [3]. Another risk factor that contributes to atresia development is maternal use of vasoconstrictive medications, which results in mesenteric blood flow interruption. Genetic predispositions also influence atresia formation- such as inherited thrombophilia, including the factor V Leiden mutation and a polymorphism of the factor VII gene, have been observed more frequently in newborns with intestinal atresia, though routine screening is not supported. These multifaceted risk factors underscore the complex etiology of ileal atresia [4].

A history and physical examination are necessary to diagnose or rule out jejunoileal atresia. The important gestational factors to be considered when jejunoileal atresia is being evaluated are family history, illicit drug use during pregnancy, gestational age at birth, complications during pregnancy, polyhydramnios, dilated bowel on ultrasound, and congenital anomalies such as Down syndrome. Because our patient presented with Down syndrome, it was imperative to further investigate his symptoms. In addition to gestational history, the documentation of perinatal history is crucial, encompassing details about delivery complications, feeding patterns, the onset of abdominal distension, and the nature of emesis (bilious or non-bilious). Additionally, noting the timing of the meconium passage is essential. Collectively, this patient not only failed to pass meconium until after 36 hours of life, but he also presented with multiple episodes of bilious emesis and abdominal distention. A comprehensive physical examination should cover an overall assessment of illness severity, abdominal distension, signs of peritonitis, respiratory issues due to aspiration or diaphragm-related concerns, indications of dehydration, presence of jaundice, and the identification of congenital anomalies such as Down syndrome, congenital heart disease, and anorectal malformations.

The mode of diagnosis of intestinal atresia depends on the course of time. Prenatally, jejunoileal atresia is diagnosed via ultrasound. On ultrasound, jejunoileal atresia presents as polyhydramnios, ascites, dilated bowel loops, and enhanced bowel echogenicity. Postnatally, JIA is diagnosed via clinical signs and symptoms and radiographic examination. Patients typically present with abdominal distention, delayed passage of meconium, and bilious emesis, as was seen in our patient. Utilizing plain X-rays with swallowed air as a contrast is a valuable diagnostic approach for examining the abdominal region. The presence of intraperitoneal calcification may suggest prenatal bowel perforation or meconium peritonitis. Malrotation is ruled out through an upper gastrointestinal series contrast study. To unveil atypical colon appearances, a contrast enema is employed, revealing characteristics seen in conditions like meconium ileus or Hirschsprung disease. After completion of the enema for our patient, we were unable to rule out these conditions, prompting further evaluation.

Upon confirming or suspecting jejunoileal atresia, initial preoperative steps involve decompression using a nasogastric tube and resuscitation with fluids and electrolytes. Intravenous broad-spectrum antibiotics are administered if there's evidence of perforation or infection. The surgical approach hinges on factors such as the location of the atresia, intraoperative conditions, and remaining bowel length. Typically, the procedure entails resection of dilated and atretic bowel, followed by primary end-to-end anastomosis, and potential tapering enteroplasty of the proximal bowel. Surgical methods may be laparoscopic-assisted or open, with considerations for the surgeon's preference, patient presentation, and anatomy. In cases of multiple segmental atresias, primary surgical repair aims to preserve bowel length, often with a proximal protective diversion for healing time. When atretic lesions are separated by a short segment, resection of the short atresia with primary anastomosis is recommended to minimize the number of intestinal anastomoses, assuming the patient has sufficient normal intestinal length.

Overall, the prognosis for intestinal atresia is highly favorable. Mortality in these patients occurs in infants with underlying or comorbid factors such as prematurity, respiratory distress syndrome, or other accompanying anatomical anomalies [3]. The primary anomalies contributing to morbidity and mortality are short bowel syndrome and congenital cardiac lesions. Short bowel syndrome is malabsorption resulting from extensive resection of the small bowel, which is more frequently a complication in patients presenting with duodenal atresia, particularly in those with Down syndrome. Because our patient presented with ileal atresia without any other comorbid conditions apart from his chromosomal anomaly, he was placed at a lower risk for having subsequent morbidity. Bowel function is the essential factor in determining the duration of necessary total parenteral nutrition, along with its associated risks of sepsis and liver failure. If patients are left with an insufficient quality of residual bowel, they are at increased risk of suffering from a poor quality of life and may be required to sustain TPN for extended periods. Primary complications may erupt due to post-surgical complications.

3. Conclusion

This case underscores the importance of early and thorough medical evaluation in infants with Down syndrome, as they may be

at an increased risk for associated infrequent GI anomalies. This report highlights a rare case of a male infant with Down syndrome diagnosed with ileal atresia and to the best of our knowledge is the first such documented case. In existing literature, ileal atresia is frequently discussed in conjunction with jejunoileal atresia. The manifestation of this condition as an isolated disorder is uncommon, and to date, there have been no documented cases in individuals with Down syndrome.

Currently, we present a case involving a 2-day-old male infant with Down syndrome, born at full term, who exhibited clinical symptoms of ileal atresia, including bilious emesis and a lack of meconium passage within the initial 48 hours of life. While gastrointestinal anomalies such as duodenal atresia (3.9%), anal stenosis/atresia (1.0%), and Hirschsprung disease (0.8%) are recognized as more prevalent in Down syndrome, the occurrence of ileal atresia in this population has not been previously reported [5-7].

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