Jejunal and Colonic Atresia

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Introduction

Jejunoileal atresia is a common cause of intestinal obstruction in neonates. The incidence of small bowel atresia is 3.4 per 10,000 live births in the American registry[1], 1.6 per 10,000 in the European registry[2], and 2.23 per 10,000 in the Japanese registry[3]. The incidence of colonic atresia is infrequent, and they comprises around 2 to 15% of intestinal atresias[4]. The first successful diagnosis and management of small bowel obstruction in a living infant was described by Sutton in 1889, who performed an ileostomy[5]. Louw and Barnard laid the experimental groundwork for the vascular insufficiency theory of intestinal atresia[6]. The current classification of jejunoileal atresia into five types was proposed by Martin and Zerella in 1976[7] and later modified by Grosfeld in 1979[8]. In 1955, Nixon suggested resection of the proximal distended bowel due to poor motility.

Embryology

Various etiological theories have been proposed to explain the development of intestinal atresia. Earlier hypothesis included fetal peritonitis; congenital volvulus or adhesive bands causing strangulation; mucosal degeneration due to unknown causes; intrauterine intussusception with absorption of necrotic bowel; abnormalities in intestinal nerve supply; occlusion of the mesenteric artery due to sclerosis; congenital syphilis; errors in intestinal rotation; pressure from surrounding organs; excessive atrophy and involution of the vitelline duct, and strangulation of a loop of bowel by an umbilical ring[5]. Although historically significant, most of these are now largely replaced.

Type I anomalies with membranous atresia can be explained by the vacuolisation theory. During the 8^{th} to 10^{th} week of gestation, the rapidly growing midgut becomes a solid cord due to overgrowth and proliferation of mucosal cells. Recanalisation occurs through the formation and coalescence of vacuoles, resulting in a patent lumen by the 12^{th} week. Failure of recanalisation can lead to a mucosal web or stenosis. [5]

In contrast, the other types of atresias can be explained by the vascular accident theory. During intestinal rotation (approximately at the 5^{th} to 12^{th} week of gestation), any malrotation or torsion of the intestine can cause necrosis of the affected gut, leading to large-gap atresia (Type IIIa, IIIb, and IV) with compromised blood and nerve supply to the blind ends.

During later fetal development, intrauterine intussusception may occur, causing localised necrosis and resorption of the invaginated segment, which leads to a short-gap atresia (Type II). Since the gut would have developed anatomically and functionally, there may be no pathological defects at the blind ends.

Genetic factors

Jejunoileal atresias are sporadic; however, some reports of concordance in twins suggest a potential genetic predisposition, especially in multiple atresias. Mutations in the Tetratricopeptide repeat domain-7A (*TTC7A*) gene have been reported in multiple intestinal atresia with autosomal recessive inheritance[9]. Other possible candidate genes include Integrin Alpha 2 (*ITGA2*) and Natriuretic peptide precursor (*NPPA*)[10]. Cystic fibrosis is another genetic condition which causes meconium ileus and meconium peritonitis, which can lead to intestinal atresia.

Risk factors

Maternal smoking and cocaine use were associated with intestinal atresia. Use of pseudoephedrine, which is a nasal decongestant, has also been linked to intestinal atresia.[11, 12] The common feature of these risk factors is their vasoconstrictive effect, which can lead to vascular injury and, in turn, cause atresia. Maternal glomerulonephritis has been shown in some studies, possibly due to altered placental perfusion.[11-13] Conversely, fish consumption in early pregnancy has been associated with a decreased risk of atresia, although the underlying mechanism is unclear.[14]

Associated anomalies

Associated anomalies are less common compared to duodenal atresia. This might be due to the development of atresia at a later stage of fetal life. Limited studies have reported the occurrence of various other anomalies, such as cardiac and renal anomalies, as well as concomitant duodenal or colonic atresia, but the incidence of such associations remains low.[15] In gastroschisis, intestinal atresias occur due to pathophysiological mechanisms, such as vascular compromise, volvulus, or compression of the exposed bowel, rather than as a coincidental congenital association.

Classification

The initial classification proposed by Louw in 1966 was based on presumed aetiology and morphological characteristics. They described three types. Type 1: an intraluminal diaphragm or membrane or stenosis, Type 2: blind ending segments connected by a fibrous cord, Type 3: blind ends separated by a V-shaped mesenteric defect[6].

In 1976, Martin and Zerella expanded this classification to four types. Their classification retained Type I and Type II as Louw, but multiple atresias were classified under Type III, and Type IV with apple-peel (or Christmas tree) deformity.[7] This modification has clinical utility, as higher types (Type III and IV) are associated with a poor prognosis.

Grosfeld later revised the classification in 1979, combining elements from both systems while keeping Louw's original terminology. In this widely accepted classification, Type I is an intraluminal web(Figure 1), Type II includes blind-ending bowel segments connected by a fibrous cord (Figure 2), Type IIIa involves blind-ending bowel segments separated by a mesenteric defect (Figure 3), and Type IIIb refers to apple-peel atresia, characterised by proximal jejunal atresia with the distal bowel spiralled around and supplied by a single marginal artery. Type IV describes multiple atresias (Figure 4) that produce a string-of-sausages appearance.[8]

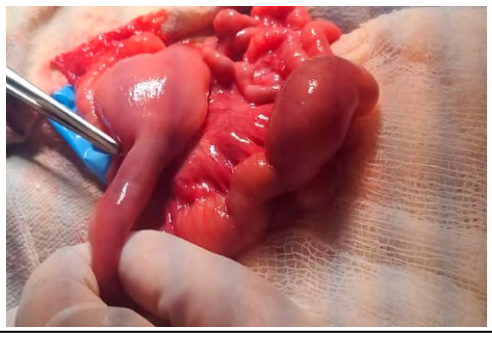

Figure 1. Type I Atresia– intraluminal web

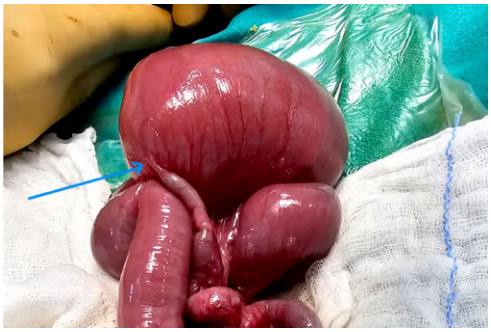

Figure 2. Type II Atresia- ends connected with a fibrous cord

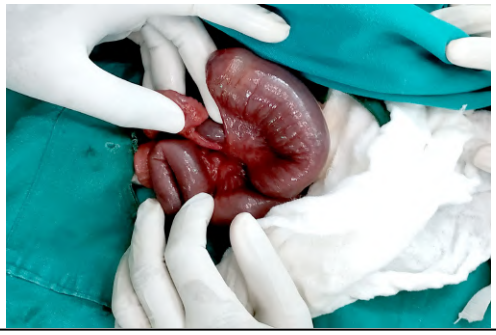

Figure 3. Type III Atresia- with mesenteric defect

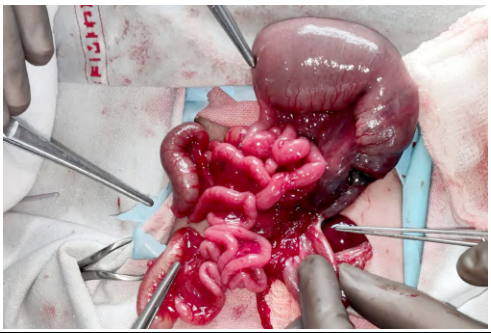

Figure 4. Type IV Atresia- Multiple atresias, along with colonic atresia, and gangrene of the proximal bowel loop

A new variant described as Type IIIc presented from a single report, in which proximal jejunal atresia is followed by a short apple peel deformity and then normal ileum[16]. In this variant, the apple-peel segment can be safely excised, and an end-to-end anastomosis between the jejunum and ileum is feasible.

For colonic atresia, the same classification system is applied (Type I – IV), although Type IIIb is not encountered in the colon.

Clinical presentation

Vomiting is the most common symptom, usually bilious and non-projectile. It can be feculent in cases of delayed presentation with bowel atresia at a more distal site. Non-passage of stool is typical, but neonates may pass a small meconium plug spontaneously or with rectal stimulation. This does not exclude the diagnosis of atresia, as bile enters the gut as early as 3 months before the atresia may develop.

The meconium may appear normal macroscopically; a microscopic examination (Farber test) might demonstrate the absence of lanugo and cornified epithelial cells.[5] These elements appear in meconium after the 5th month of gestation, when hair formation begins. The presence of lanugo may suggest a late-onset intrauterine obstruction.

Abdominal distension is another prominent symptom, which becomes more noticeable with delayed diagnosis. The more distal the level of obstruction, the more severe the distension. Prominent distended bowel loops may be visible, with or without peristalsis. The presence of redness over the abdominal wall, tenderness, and a mass may indicate bowel gangrene.

Jaundice is another clinical symptom that develops due to intestinal obstruction in neonates. The exact pathophysiology is not fully understood, but potential mechanisms may include increased enterohepatic circulation and impaired intestinal transit, leading to indirect hyperbilirubinemia.[17] However, this does not fully explain why jaundice is more pronounced with high obstruction than with low obstruction.[18] It often improves spontaneously with surgical management. Studies have shown that jaundice responds well to phototherapy.

Diagnosis

Prenatal diagnosis

The detection rate of small bowel atresia varies from 25% to 90%[19]. The diagnosis may not be precise in the second trimester due to late developmental events, as described earlier in embryology. However, the progression of signs in the third-trimester scan can be helpful. Hyper echogenicity of the bowel—meaning echogenicity equal to or greater than bone, dilated bowel loops (generally >7mm diameter; some authors use > 17mm or diameter to gestational age more than 0.49), dilated stomach, ascites, cysts, polyhydramnios, and intra-abdominal calcifications are the common sonographic findings[20, 21]. Nonetheless, differentiating between meconium ileus, atresia, and Hirschsprung's disease may not be possible with ultrasound.

Fetal MRI can identify the level of obstruction in about 90% of cases; however, the physiological hypersignal on T1 sequences due to protein-rich meconium content should be taken into account.[21]

Postnatal diagnosis

A plain X-ray of the abdomen, taken both supine and erect, is the initial radiological investigation.[22] An early X-ray may appear non-specific, as gas reaches the rectum only by 12 hours of life, and this delay can be seen in preterm infants. A supine film demonstrates bowel gas patterns, such as the double or triple-bubble signs, which indicate whether the obstruction is high or low. An erect film (if feasible) can reveal dilated bowel loops with air-fluid levels or air under the diaphragm. If an erect position is not possible, a lateral decubitus or cross-table X-ray can be helpful [23]. A plain X-ray might suggest an obstruction, but it is not specific. Differentiating between small bowel obstruction and large bowel obstruction is difficult in neonates.[24]

A contrast enema can help when the diagnosis is uncertain. A rectal contrast study can identify an unused colon and transition zone in Hirschsprung's disease, colonic atresias and the presence of meconium pellets ("string of beads") at the ileocecal region in meconium ileus. Furthermore, contrast studies can evacuate distal bowel contents, thereby facilitating decompression before anastomosis. The decision to perform a contrast study must be individualised.

Management

The initial management should focus on correcting or preventing hypothermia, hypovolemia, hypoglycaemia, and hypoxemia.[25] The baby should be kept in a pre-warmed incubator to maintain temperature, and intravenous fluids should be warmed before administration. Urine output is an essential indicator of hydration status.

Significant dehydration may be present in out born babies; a bolus of 10-20 mL/kg of Ringer's lactate solution may be required. Vomiting and sequestration of fluid in dilated bowel loops can cause third-space loss and electrolyte imbalance, particularly hypokalemia. Placing a nasogastric or orogastric tube can help decompress the bowel, prevent aspiration during vomiting, and prevent progressive bowel distension that can lead to bowel gangrene. NG output can be replaced volume-for-volume with Ringer's lactate every 8 to 12 hours. Raising lactate levels and metabolic acidosis may indicate bowel gangrene. Blood products, including packed cells and fresh frozen plasma, should be arranged. Vitamin K1 and broad-spectrum IV antibiotics should be administered before surgery.

There are no specific recommendations for the timing of surgery; it depends entirely on the baby's hemodynamic stability. For inborn babies with an antenatally suspected intestinal atresia, waiting 24 to 48 hours can help in evaluating the level of obstruction and assessing any associated

anomalies before surgery, provided there is no suspicion of malrotation, volvulus or bowel gangrene. O'Neill, et al. [5] reported that gangrene of the proximal bowel can occur as early as 48hours. Colonic atresia carries a high chance of gangrene with a competent ileocecal junction.

Abdominal access can be obtained via a laparotomy, performed through a right upper quadrant transverse incision or a supraumbilical incision.[26] Laparoscopy-assisted surgery [27] is an alternative, which involves identifying atretic ends, which are then brought out through an enlarged umbilical incision for extracorporeal anastomosis, with or without staplers.

Bowel loops are examined to determine the type of anomaly, discrepancies in bowel ends, gangrene, and any associated anomalies. The distal bowel should be evaluated for additional atresia or stenosis. This can be done by passing a feeding tube or IV cannula into the distal atretic end and irrigating with saline until the rectum if a contrast enema was not performed before surgery, or until the ileocolic junction if there is sufficient evidence of colon patency from a contrast enema.

For type I stenosis or membranous obstruction, an enterotomy and web excision are sufficient. When narrowing occurs at the membrane level, a Y-V enteroplasty can be performed along with web excision.[28] For a type II or short-gap mesenteric defect, the management includes excision of the atretic ends and end-to-end-back anastomosis. End-to-back anastomosis involves connecting the end of the proximal bowel with the back of the distal bowel (back wall or the antimesenteric side of the bowel opened to match the diameter of the proximal bowel). This helps to overcome luminal discrepancy, and a wider anastomosis reduces the risk of stricture. A side-to-side anastomosis is not advisable, as it increases the risk of creating a blind loop and of retaining poorly motile segments of the proximal bowel. It is important to excise the atretic segments, as advocated by Louw et al.[6], because these ends will have ineffective peristalsis. For type IIIa with a large mesenteric gap, the choice of surgery depends on the length of remaining bowel and the degree of proximal segment dilation. The length of the segment to be excised varies; if sufficient bowel length is available, the bulbous part of the proximal atretic end can be excised until peristalsis is observed on tapping and the bowel has a relatively better calibre. Saha et al. [29] examined the histopathology of the atretic ends and showed that segments proximal to 5 to 8 cm have a better density of interstitial cells of Cajal and normal muscle architecture. If there is insufficient bowel, tapering enteroplasty or plication can be considered.[30, 31]

Type IIIb (apple peel atresia) and Type IV, involving multiple atresias, present management challenges. Since these cases often have a relatively short bowel length, the distal bowel in apple peel atresia is small-calibre and twisted around a single artery, risking compromised blood supply. Infusing saline to evaluate distal atresias can slightly enlarge the calibre before performing anastomosis. The adhesive bands attached to the distal bowel should be carefully released. A primary end-to-back anastomosis is preferred when feasible. For multiple atresias, multiple anastomoses may be performed if the baby is hemodynamically stable and tolerates a longer surgery. Otherwise, a stoma can be created to ensure sufficient bowel length and prevent short gut

syndrome. Other alternative procedures include the Bishop-Koop or Santulli procedures, especially for Type IIIb and IV cases, those with significant luminal discrepancy, and those with meconium peritonitis.[32]

In all these cases, if total parenteral nutrition (TPN) is unavailable or limited, a trans-anastomotic feeding access can be established via gastrostomy.[33] However, this procedure has its own complications, and some studies have shown a delay in oral feeding with this method. If TPN is available, it is generally preferable due to its better nutritional profile, particularly in prolonged ileus and short gut risk cases.

Colonic atresias, which are most common in the right colon, are usually managed by creating a stoma initially, with closure scheduled later. This approach is mainly due to the discrepancy of the ends, which can lead to anastomotic leaks. Some studies have performed primary anastomosis and reported successful outcomes.[4]

In atresias associated with gastroschisis, proximal lesions are better managed with a delayed primary repair, as bowel oedema will hamper identification and repair. In contrast, distal atresias are usually apparent and often complicated by infarction or perforation; therefore, making a stoma initially and delaying restoration of bowel continuity until bowel oedema subsides is a better option.

Drains are optional; if placed, they may help detect a leak and distinguish it from ileus, which is expected in complex atresias.

Postoperative management

Frequent monitoring of electrolytes and acid-base balance is necessary. Replace the NG output promptly. If a transanastomotic feeding access is established, enteral feeding is usually initiated by postoperative day 4 to 5, or after the baby passes stools without abdominal distension. If a transanastomotic feeding tube is not placed, or in complex cases such as Type IIIb or Type IV, bowel peristalsis might be delayed by 2 to 3 weeks.

Minor anastomotic leaks can be managed conservatively with bowel rest, antibiotics, drainage and TPN if the baby is hemodynamically stable. Major leaks require reoperation with redo anastomosis or stoma with feeding access. Prolonged ileus is common in complex atresias, and serial clinical assessment is required. A contrast follow-through X-ray might be necessary to distinguish adhesive bowel obstruction, ileus, and leak. Some studies reported a mean TPN duration of 2 months in Type IIIb cases.[34]

Long-term follow-up

The prognosis for these children depends on the length and quality of the remaining bowel. Children with short bowel syndrome will be dependent on TPN or may require bowel lengthening

procedures. Malabsorption, growth failure and chronic malnutrition can lead to impaired immune function and developmental delay. There is no uniform data available on mortality and morbidity rates across different types of anomalies; however, Types I and II generally have better outcomes as they have sufficient bowel length. Children with Type III and IV typically have poorer outcomes. These children are more prone to developing sepsis and pneumonia due to prolonged hospitalisation and dependence on TPN.

Future directions and research

Recent animal studies have explored the use of biomarkers such as intestinal fatty acid-binding protein (I-FABP) and Trefoil factor 3 (TFF3)[35]. These may not aid early detection of atresia; their potential role lies in early prediction of ischemic injury[36]. These are surrogate markers of transmural necrosis and may help in assessing the doubtful bowel segments left in situ for a second-look laparotomy.

The emergence of three-germ-layer organoids has helped in understanding embryological failures leading to atresia[37]. Researchers used these models to identify specific mutations and abnormalities in signalling pathways in duodenal atresia[38]. They might also help identify which segment of the bowel is involved in an anastomosis that leads to dysmotility after a successful anastomosis. The most promising aspect of these organoids is the development of patient-specific intestinal grafts. In cases of multiple atresia or extensive bowel loss due to gangrene leading to short bowel syndrome, these patient-specific intestinal grafts will be extremely helpful.

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