Colonic and Rectal Atresias

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Abstract

Colonic and rectal atresias are very rare. Both have been considered to be the result of insufficient blood supply during intrauterine life. Rectal atresia is included in classifications of anorectal malformations. Colonic and rectal atresia both present as neonatal bowel obstruction. The diagnosis of colonic atresia is usually suspected on plain abdominal radiographs. A contrast enema is useful to show a microcolon. Rectal atresia is suspected in neonates with bowel obstruction, where it is impossible to pass a catheter through the rectum, although the anus is normal. Colonic atresia is often operated with resection of dilated colon and primary anastomosis. In complicated cases, it is common to open a colostomy in the neonate and do the anastomosis later. There are several procedures for rectal atresias. Many investigators open a colostomy in the neonate and reconstruct the rectum through a posterior

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sagittal approach at a later stage. Long-term functional outcome is usually favorable in patients with colonic as well as rectal atresia.

**Keywords**
Colonic atresia · Rectal atresia · Primary anastomosis · Posterior sagittal approach · Outcome

**Colonic Atresia**

**Introduction**

Binninger was the first to describe colonic atresia in 1673 (Evans 1951). The first survivor was reported in 1922, when Gaub (1922) opened a diverting colostomy in a child with an atresia of the sigmoid colon. Potts (1947) successfully performed a primary anastomosis in a neonate with an atresia of the transverse colon in 1947.

Atresia of the colon is a rare cause of bowel obstruction in the neonate. The incidence of colonic atresia in live births has been difficult to ascertain, but an incidence of approximately one in 20,000 live births has been considered to be realistic based on the experience in major pediatric surgical centers (Philippart 1986). In the Northwest of England, isolated colonic atresia has been reported to occur in one in 66,000 live births (Davenport et al. 1990). Other investigators have reported that colonic atresias account for 1.8–10.5% of the total bowel atresias (Freeman 1966; Benson et al. 1968), the incidence of which has been estimated to be one in 1500 to one in 20,000 live births (Evans 1951; Webb and Wangensteen 1931). Colonic stenosis is extremely rare.

Except for stenosis, three different types of intrinsic occlusion have been distinguished (Louw 1964; Bland Sutton 1889):

1. Type I atresia or a membrane (Fig. 1a)
2. Type II atresia with blind ends of bowel joined together by a cord-like remnant of bowel, with or without a gap in the mesentery (Fig. 1b)

3. Type III atresia with separated blind ends of bowel and a gap in the mesentery (Fig. 1c)

Furthermore, a hereditary form with multiple atresias of the gastrointestinal tract has been described, suggested to be of nonvascular origin (Guttman et al. 1973; Puri and Fujimoto 1988). Type III atresia appears to be the most common type proximal to the splenic flexure, whereas type I and II are more common in atresias distal to the splenic flexure (Powell and Raffensperger 1982; Boles et al. 1976). In a literature review, it was reported that type III occurred in 60.4% of all...
cases (Etensel et al. 2005). Most series show an even distribution between atresias proximal and distal to the splenic flexure (Freeman 1966; Benson et al. 1968; Peck et al. 1963; Coran and Eraklis 1969).

**Etiology**

Colonic atresia is probably the result of intrauterine vascular insufficiency. The finding of bile, squamous epithelium, and hair in the bowel distal to the atresia supports the hypothesis that the vascular accident occurs late in development (Louw 1964). Several pathological conditions may result in compromised blood supply to the bowel, such as intussusception, volvulus, herniation, tight gastroschisis, and embolic or thrombotic events. It appears likely that focal resorption of the sterile gut occurs after ischemic necrosis. Animal experiments have been performed in which the blood supply was interrupted to different parts of the small intestine or colon, thus inducing various types of atresias. These experiments confirm the etiologic role of intrauterine vascular occlusion (Louw 1964; Barnard and Louw 1956; Louw and Barnard 1955).

Colonic atresia has been reported in monozygotic twins (Kim et al. 2000). Benawraet et al. (1981) reported three cases occurring in first-degree relatives of a family. Fairbanks et al. (2005) has shown that the absence of fibroblast growth factor 10 (Fgf10) or its receptor fibroblast growth factor receptor 2b (Fgfr2b) results in colonic atresia in a mouse model, despite normal mesenteric vascular development. These findings suggest that genetic factors may play a role in the pathogenesis of colonic atresia.

**Presentation**

Neonates with colonic atresia present with symptoms of distal bowel obstruction. Abdominal distension is usually present at birth but otherwise develops over the first 24–48 h of life. Bile-stained vomiting is very common but is not always an early symptom. Failure to pass meconium is the rule, and neonates that do not pass meconium within the first 24 h of life should be considered for further investigations. On examination, the abdomen is distended and often slightly tender, sometimes with visible bowel loops. In those who have an abdominal wall defect, associated atresias should always be suspected.

Colonic atresia is associated with abdominal wall defects, such as gastrochisis, cloacal exstrophy, and more rarely omphalocele, which complicates the management of these patients (Philippart 1986; Boles et al. 1976; El-Asmar et al. 2016). Boles et al. (1976) found that four of their 11 patients had gastrochisis. In the series reported by Philippart (1986), 22 of 36 patients with colonic atresia had no associated anomalies, whereas six had cloacal exstrophy, and three had other abdominal wall defects. Five of the 36 patients had jejunal atresia associated with the colonic atresia. Rarely, colonic atresia has been reported to occur concomitantly with imperforate anus (Benson et al. 1968). Malrotation has also been reported to be a common associated anomaly (Etensel et al. 2005). One important associated anomaly is Hirschsprung’s disease, which has been reported in a few cases (El-Asmar et al. 2016). Although the colonic atresia was diagnosed at birth in these patients, there was a considerable delay in diagnosing the associated aganglionosis. It is therefore recommended that resected bowel is examined for Hirschsprung’s disease (Akgur et al. 1998). Rectal suction biopsies are suggested in patients that do not gain normal bowel function postoperatively (Kim et al. 1995). Some authors recommend that rectal suction biopsies should be routinely taken in all patients with colonic atresia (Etensel et al. 2005; Lauwers et al. 2006). Isolated colonic atresia is sometimes associated with skeletal anomalies such as syndactyly, polydactyly, absent radius, and clubfoot (Philippart 1986). Furthermore, colonic atresia has been reported in association with eye anomalies, such as exophthalmos and optic nerve hypoplasia (Powell and Raffensperger 1982). In the series reported
by Davenport et al. (1990), one patient had trisomy 18 and esophageal atresia. The fact that chromosomal abnormalities do occur in patients with colonic atresia makes it reasonable to recommend chromosomal analysis, at least in those patients who have other associated anomalies.

**Diagnosis**

Prenatal diagnosis of colonic atresia has been reported. However, prenatally detected colonic dilatation may also be the result of Hirschsprung’s disease or anorectal malformations (Anderson et al. 1993).

Plain radiographs show a distal bowel obstruction with multiple dilated loops with air-fluid levels. A large right-sided loop, corresponding to the proximal dilated colon, has been considered characteristic in patients with colonic atresia (Fig. 2a) (Davenport et al. 1990). The level of obstruction is confirmed by a contrast enema, which reveals the distal microcolon and incomplete filling of the colon (Fig. 2b). Pneumoperitoneum, indicating colonic perforation, is not rare and has been reported in approximately 10% of the cases (Philippart 1986).

**Management**

**Preoperative**

Correction of fluid and electrolyte abnormalities is started as soon as bowel obstruction is suspected. The gastrointestinal tract is decompressed with a nasogastric tube. Prophylactic antibiotics are administered. The neonate should be in a stable condition before general anesthesia and the operation are started.

**Operative**

The two therapeutic options available are primary resection with anastomosis and colostomy with anastomosis at a later stage. Traditionally, many authors distinguished between the management of colonic atresias distal and proximal to the splenic flexure. Atresias proximal to the splenic flexure were treated with primary resection and anastomosis, whereas the distal atresias were treated with primary colostomy and delayed establishment of the gastrointestinal continuity (Philippart 1986; Benson et al. 1968; Powell and Raffensperger 1982; Coran and Eraklis 1969; Defore et al. 1976). More recently, it has been suggested that staged repair should be undertaken in complex cases with, for instance, questionable

![Fig. 2](a) Plain abdominal radiographs often show the hugely dilated bowel segment proximal to the atresia. (b) A contrast enema is diagnostic of a colonic atresia showing a microcolon and incomplete colonic filling.
bowel viability, colonic perforation, and peritonitis and in patients with concomitant abdominal wall defects. On the other hand, in uncomplicated cases, resection and primary anastomosis has been proposed to be the method of choice for atresias at all levels of the colon (Arca and Oldham 2012). There is no evidence that this later approach increases the mortality or complication rate (Davenport et al. 1990), although the anastomosis may be technically difficult because of the large discrepancy between the diameters of the proximal and distal bowel (Watts et al. 2003).

The abdomen is opened through a transverse incision a finger diameter above the umbilicus and to the right. The incision may be extended as required. Cautery is used to divide the muscle layers of the abdominal wall, and the umbilical vein is ligated and divided. The site and type of atresia is assessed (Fig. 3). It is extremely important that additional atresias are excluded. The patency of the distal colon must always be tested by, for instance, injection of saline. In those with type I atresias, the bowel adjacent to the atresia is resected, and a primary anastomosis is performed. In patients with type II and III atresias, with adequate bowel length, the excessively dilated proximal bowel should also be resected (Fig. 4a). A few centimeters of the distal narrow bowel are resected. The mesenteric vessels are divided close to the bowel wall to preserve the blood supply to the adjacent bowel. The distal bowel is incised along the antimesenteric border to match the luminal diameters of the two portions of bowel to be joined (Reprinted with permission from Wester T. Colonic and Rectal Atresias. In Puri P (ed). Newborn Surgery 3rd ed. Hodder Arnold, London. 2011 pp 505–511, Fig. 53.3)

Fig. 3 The proximal colon is hugely dilated (arrow a), while the distal colon is very small (arrow b)

Postoperative
During the first postoperative days, parenteral nutrition is administered. Feeding can be started when the baby is well and the gastric aspirates have decreased. In cases with a primary anastomosis, it usually takes a few days before the neonate starts to pass stools. If a colostomy has been fashioned, the parents are instructed how to take care of the stoma. Usually, the colostomy is closed at 2–3 months of age.
Complications and Long-Term Results

Many factors have led to an improvement in the results of patients with colonic atresia, including early postnatal diagnosis, improved neonatal intensive care and anesthesia, and more efficient transport facilities. Today, mortality related to the colonic atresia or its treatment is rare. In the series reported by Davenport et al. (1990), no deaths occurred in the patients that underwent surgery, although one patient, who was never operated on, died of associated abnormalities. The mortality rate in earlier series varied from 9% to 33%, in many cases as a result of associated anomalies but also attributable to late diagnosis, nutritional deficiencies, infectious complications, and technical errors (Freeman 1966; Benson et al. 1968; Powell and Raffensperger 1982; Boles et al. 1976; Coran and Eraklis 1969). Etensel et al. (2005) recently reported a lethal outcome in 27% of the cases collected for a literature review.

Powell et al. (Powell and Raffensperger 1982) reported 15 postoperative complications in 19 patients. Problems related to the colostomy were encountered in three of 11 patients treated with colostomy and delayed anastomosis, whereas anastomotic strictures were seen in six of the 19 patients. Boles et al. (1976) reported significant complications in four of 11 cases. The use of contemporary principles of neonatal surgery has, however, reduced the morbidity rate, and Davenport et al. (1990) reported recovery without complications.

Rectal Atresia

Introduction

Rectal atresia has been classified under the rare types of anorectal malformations (ARM) in the Krickenbeck classification constituting only 1–2% of ARM cases (Sharma and Gupta 2017). Rectal atresia is characterized by the presence of a normally developed anus and sphincter muscles with a proximal blind and dilated rectum, which wends at or above the psoococcygeal line. Sharma and Gupta (Sharma and Gupta 2017) have classified rectal atresia into five types: Type I: Rectal stenosis: (A) Intramural (B) Web with a hole. Type II: Rectal atresia with a septal defect. Type III: Rectal atresia with a fibrous cord between two atretic ends. Type IV: Rectal atresia with a gap (Fig. 6) Type V: Multiple: (A) rectal atresia with stenosis (B) multiple rectal atresia and (C) thickened Houston’s valves/multiple rectal stenosis.
**Etiology**

The exact embryogenic process underlying this anomaly is not known, but the following theories have been postulated.

1. Embryological theory: rectal atresia develops due to a vascular accident that occurs at a window in time between 13 and 14 weeks of gestation.
2. Genetic theory: there is high incidence of these anomalies among consanguineous marriages, especially in South India. This supports the genetic association of the condition.
3. Infective theory: this theory was supported by Magnus (1968) and she believed that intrauterine infection causing thrombosis of the vessels could lead to acquired atresia of the already formed rectum.

Due to the pattern of geographic distribution noted with this anomaly, a racial or genetic defect may be much more important as the possible etiological factor.

**Presentation**

Neonates with rectal atresia present with distal bowel obstruction comprising abdominal distension and failure to pass meconium. The perineum and anal canal are normal, and the diagnosis may therefore easily be delayed. The atresia is usually located 1–3 cm above the dentate line.

The incidence of associated anomalies in patients with rectal atresia has been considered to be extremely low (Peña 1996). In the series reported by Dorairajan (1988), associated anomalies were found in 2% of the 147 cases. No significant abnormalities were found in the urinary tract. Patients with rectal atresia usually have a normal perineum and a normal sacrum. Rectal atresia occurs in patients with multiple atresias of the bowel (Magnus 1968). Two of Dorairajan’s (1988) patients had ileal atresia, and one had multiple small bowel atresias. Patients with rectal atresia and particularly rectal stenosis may have an associated presacral mass, and it has been recommended that they should undergo MRI like other patients with anorectal malformations. Presacral mass such as presacral teratoma or anterior sacral meningocele is the most commonly reported association with rectal atresia/rectal stenosis, 29% patients in Penas’s series (Hamrick et al. 2012). Sharma and Gupta (Sharma and Gupta 2017) reported 10 cases of rectal atresia/rectal stenosis over a 15 year period. Median age at presentation was 1 month (1 day–96 months). Six patients including all five cases of rectal atresia underwent colostomy at birth.

**Diagnosis**

The diagnosis of rectal atresia can be challenging as the patients show a normal anus. The inability to pass a thermometer or a rectal tube up the anal canal should raise the suspicion of rectal atresia. After a colostomy has been opened, a contrast study with simultaneous injection of contrast material through the colostomy into the rectal pouch and the anal canal clearly outlines the anatomy of the anomaly (Fig. 7).
Management

Various procedures have been described to treat rectal atresia and rectal stenosis, many of which have now become obsolete such as abdominoperineal and sacroperineal pullthrough operations (Sharma and Gupta 2017). Most commonly used procedures now a days for rectal atresia and rectal stenosis include posterior saggital anorectoplasty and transanal pull-through operation. Upadhyaya (Upadhyaya 1996) recommended transanal end to end recto-rectal anastomosis. The advantage of this technique is that the luminal continuity is restored without causing damage to functional anatomy of the region.

Complications and Long-Term Results

In patients with rectal atresia, the anal canal, sacrum, and sphincteric mechanisms are virtually normal. Therefore, the prognosis with respect to functional outcome is favorable. Although the number of cases reported is very limited, the outcome in patients with rectal atresia or stenosis treated through a posterior sagittal approach is excellent. Peña (1995) reported voluntary bowel movements with total continence and without soiling in a series of five cases. However, two of the five patients had constipation. The same group showed, in a more recent series of 17 patients, that all 12 patients older than 3 years had voluntary bowel movements and were clean between bowel movements. Five (29%) of the 17 patients had constipation that required laxatives. The authors report that complications occurred in three patients; one rectovaginal fistula and two presacral abscesses (Hamrick et al. 2012). Constipation has been reported to occur frequently after other procedures used to treat rectal atresia or stenosis as well (Zia-ul-Miraj Ahmed et al. 1995). Upadhyaya (1990) reported an uneventful recovery and normal continence in two patients treated with his method. Dorairajan (1988) followed up 37 of 60 patients that were treated with sacroperineal pull-through operations and who had their colostomy closed. The outcome was excellent in 20% of the patients, whereas 65% had occasional soiling at night, and 15% had soiling also in daytime. The mortality rate in this series was 35%. A recent study reported normal bowel control at 3–5 years of age following transanal endorectal pull-through operation for rectal atresia (Gieballa et al. 2018).

Conclusion and Future Directions

Colonic and rectal atresias are both very rare conditions. The long-term results appear to be good, although there is limited data on the outcome in adulthood in patients with colonic and rectal atresia. For colonic atresia, the surgical options are mainly resection and primary anastomosis or colostomy with delayed anastomosis. For rectal atresias, several techniques have been described. There is little evidence to show that one technique is better than another. It is, also with multicenter studies, difficult to get a sample size that is large enough to compare the methods.
Cross-References

- Anorectal Anomalies
- Embryology of Congenital Malformations
- Gastrochisis
- Jejuno-Ileal Atresia and Stenosis
- Omphalocoele
- The Bladder Exstrophy, Epispadias, Cloacal Exstrophy
- The Epidemiology of Birth Defects
- Urogenital Sinus and Cloacal Anomalies

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