Section: Paediatric Surgery

Case Series

NEONATAL TOTAL COLONIC AGANGLIONOSIS: A CASE SERIES

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ABSTRACT

Total colonic aganglionosis (TCA) is a rare and severe variant of Hirschsprung disease characterised by the absence of ganglion cells throughout the colon, often extending into the distal small intestine, and results in high-risk early morbidity. It usually presents in the neonatal period with features of intestinal obstruction such as failure to pass meconium, progressive abdominal distension, and bilious vomiting. Atypical manifestations, including spontaneous intestinal perforation, meconium ileus–like presentation, delayed obstruction after initial meconium passage, and association with syndromic conditions, may occur. Diagnosis is challenging because radiological findings may show a microcolon without a clear transition zone, and rectal suction biopsy can be inconclusive. Multiple intraoperative full-thickness biopsies from different bowel levels are essential to confirm the diagnosis and determine the proximal level of ganglionated bowel. Current expert recommendations suggest a staged surgical approach, with initial diversion ileostomy for decompression, nutritional stabilisation, and histopathological mapping, followed by definitive pull-through at a later stage. Despite advances in management, TCA is associated with higher rates of enterocolitis, perforation, dysmotility, short bowel syndrome, and prolonged dependence on parenteral nutrition compared with short-segment disease. This case series describes six neonates with TCA, highlighting the spectrum of clinical presentation, diagnostic challenges, and initial surgical management in our centre.

INTRODUCTION

Total colonic aganglionosis (TCA) is a rare and severe form of Hirschsprung disease, where ganglion cells are absent throughout the entire colon and can also affect smaller parts of the bowel. It accounts for 2–13% of all Hirschsprung disease cases and affects about 1 in 50,000 live births.[1,2] Compared to short-segment Hirschsprung disease, TCA leads to more severe and earlier symptoms because a larger part of the intestine lacks nerve cells.[3] Newborns with TCA most often show signs of intestinal blockage soon after birth. Common symptoms include failure to pass meconium in the first 48 hours, abdominal distension, bilious vomiting, and feeding issues.[4,5] In some cases, the condition appears with unusual signs such as bowel perforation leading to pneumoperitoneum, meconium ileus-like picture with pellets in the terminal ileum, or even initial passage of meconium after stimulation before later obstruction develops.[6,7] TCA can also associate with other conditions, like Shah-Waardenburg syndrome, shown by features such as a white forelock and heterochromia.[8] Recent expert consensus and reviews emphasise that TCA requires a structured, stepwise approach from early diagnosis to surgical management and long-term follow-up, and is associated with poorer outcomes compared to shorter-segment disease.[1,2] Initial management includes diversion ileostomy for decompression and multiple biopsies to confirm the diagnosis. Definitive treatment involves removal of the aganglionic bowel and a staged pull-through procedure, often with a protective stoma to reduce complications such as enterocolitis and short bowel syndrome.[1] In this report, we describe six newborns diagnosed with TCA at our centre. We aim to highlight the different ways this condition can present and explain how we diagnosed and initially managed these cases in our clinical setting.

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CASE PRESENTATION

Case 1

A 2-day-old moderate preterm male neonate (35 weeks of gestation) presented with abdominal distension since birth, tachypnea, bilious vomiting, and failure to pass meconium. An antenatal ultrasound had shown polyhydramnios. On general examination, the baby was dehydrated with a poor cry and activity. Abdominal examination revealed gross distension with visible dilated bowel loops and sluggish bowel sounds. Rectal wash produced a clear aspirate with no meconium. Abdominal X-ray showed dilated bowel loops, pellet-like structures in the terminal ileum, and an absent rectal gas shadow [Figure 1]. Intraoperative findings revealed that the small bowel was dilated up to the terminal ileum and loaded with meconium pellets. Microcolon was present. The initial intraoperative impression was meconium ileus [Figure 2]. A diversion ileostomy was performed, and multiple seromuscular biopsies were taken from different levels of the colon. Histopathology examination revealed the absence of ganglion cells in all colonic biopsy segments, with ganglion cells present at the ileostomy site, confirming TCA.

Figure 1: Abdominal X-ray showing dilated bowel loops with pellet-like opacities in the terminal ileum with absent rectal gas shadow

Figure 2: Intra OP photograph showing dilated small bowel loaded with meconium pellets and microcolon.

Case 2

A 1-day-old term male neonate (37 weeks of gestation) presented with abdominal distension and failure to pass meconium. An antenatal ultrasound had shown polyhydramnios. On general examination, the baby had a white forelock and heterochromia [Figure 3]. The abdomen was distended with visible bowel loops and sluggish bowel sounds. Rectal wash produced a clear aspirate with no meconium. Abdominal X-ray revealed dilated small bowel loops and microcolon [Figure 4-6]. In intra-operative findings, the small bowel was dilated up to the terminal ileum with a transition zone, and a microcolon was present [Figure 7]. Diversion ileostomy was performed, and multiple biopsies were taken from different levels of the colon. Histopathology examination showed absent ganglion cells in all colonic biopsy segments except the ileostomy site, confirming TCA. The presence of a white forelock and heterochromia, together with TCA, confirmed the diagnosis of Shah-Waardenburg syndrome.

Figure 3: Clinical photograph of the neonate demonstrating white forelock and heterochromia, suggestive of associated Shah-Waardenburg syndrome.

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Figure 4: Abdominal X-ray – frontal view

Figure 5: Abdominal X-ray – oblique view

Figure 6: Abdominal X-ray – lateral view.

Figure 7: Intraoperative photograph showing dilated small bowel with transition zone and microcolon.

Case 3

A 3-day-old preterm female neonate (33 weeks of gestation) presented with abdominal distension starting from the second day of life and failed to pass meconium. On general examination, the baby was dehydrated. Abdominal examination showed gross distension with visible bowel loops, absent bowel sounds, guarding, and rigidity. Rectal wash produced a clear aspirate with no meconium. Abdominal X-ray revealed pneumoperitoneum [Figure 8]. Intraoperatively, the small bowel was dilated up to the terminal ileum, and a microcolon was present with a perforation at the rectosigmoid junction. Primary closure of the perforation was performed, followed by diversion ileostomy and multiple biopsies from different levels of the colon. Histopathology examination confirmed absent ganglion cells in all colonic biopsy segments, with preserved ganglion cells at the ileostomy site, confirming the diagnosis of TCA.

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Figure 8: Abdominal X-ray demonstrating free intraperitoneal air consistent with pneumoperitoneum.

Case 4

A 2-day-old moderate preterm female neonate (32 weeks of gestation) presented with abdominal distension since birth, bilious vomiting, and failure to pass meconium. On general examination, the baby had mild dehydration. Abdominal examination showed distension with visible bowel loops and absent bowel sounds. Per rectal examination with rectal wash produced a clear aspirate with no meconium. Contrast enema study revealed the classic question-mark sign, along with dilated small bowel up to the terminal ileum and microcolon [Figure 9]. Intraoperatively, the small bowel was dilated up to the terminal ileum, and a microcolon was confirmed. Diversion ileostomy was performed, and multiple biopsies were taken from different levels of the colon. Histopathology examination showed absent ganglion cells in all colonic biopsy segments, with preserved ganglion cells at the ileostomy site, resulting the diagnosis of TCA.

Figure 9: Contrast enema showing the classic question-mark sign with dilated small bowel loops, transition in the terminal ileum, and microcolon.

Figure 10: Contrast enema showing microcolon with dilated small bowel loops up to the terminal ileum.

Case 5

A 5-day-old term female neonate presented with on-and-off bilious vomiting. The baby had passed meconium with rectal stimulation on the first day of life. On general examination, the abdomen was soft and mildly distended with active bowel sounds. The rectal examination showed meconium staining. During the hospital stay, the baby developed bilious vomiting and abdominal distension. The baby was kept nil per oral (NPO), started on intravenous fluids, and given N-acetylcysteine. Initially, she passed stools, and the distension improved. After 2 days, oral feeds were restarted. However, after 3–4 days of oral feeds, abdominal distension and bilious vomiting

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recurred. Abdominal X-ray showed dilated bowel loops. Contrast enema study revealed a microcolon [Figure 10]. Intraoperatively, the small bowel was dilated up to the terminal ileum with a microcolon. Diversion ileostomy was performed, and multiple biopsies were taken from different levels of the colon. Histopathology examination confirmed absent ganglion cells in all colonic biopsy segments, with preserved ganglion cells at the ileostomy site, resulting in the diagnosis of TCA.

Case 6

A 1-day-old term male neonate presented with abdominal distension, on-and-off bilious vomiting, and failure to pass meconium. Antenatal scans had shown polyhydramnios. On general examination, the baby was dehydrated with poor cry and activity. Abdominal examination revealed distension, absent bowel sounds, guarding, and rigidity. Per rectal examination showed no meconium staining. Abdominal X-ray demonstrated pneumoperitoneum [Figure 11]. Intraoperatively, the small bowel was dilated up to the terminal ileum with a microcolon and a perforation located 10 cm proximal to the ileocecal junction. Diversion ileostomy was performed at the site of perforation, with multiple biopsies from different levels of the colon. Histopathology examination confirmed absent ganglion cells in all colonic biopsy segments, with preserved ganglion cells at the ileostomy site, resulting diagnosis of TCA.

Figure 11: Abdominal X-ray showing free intraperitoneal air consistent with pneumoperitoneum.

Table 1: Summary of all cases

Case	Key Features	Antenatal	General Exam	Examination	Imaging	Surgical Findings
1 (2 d, preterm, AGA, M)	Abd distension after feeds, bilious vomiting, no meconium	Polyhydramnios	Poor cry & activity, tachypnea	Distended abdomen, dilated loops, sluggish BS, PR: no meconium, rectal wash clear	Dilated bowel loops, absent pelvic rectal gas	Terminal ileum with meconium pellets (meconium ileus), microcolon; diversion ileostomy + multiple biopsies; HPE: absent ganglion cells
2 (1 d, term, SGA, M)	Abd distension, no meconium	Polyhydramnios	Good cry & activity	Distended abdomen, visible loops, sluggish BS, PR: no meconium, wash clear	Dilated small bowel	Microcolon; diversion ileostomy + multiple biopsies; HPE: absent ganglion cells
3 (3 d, preterm, AGA, F)	Abd distension, no meconium	Normal	Poor cry & activity	Distended abdomen, absent BS, guarding & rigidity, PR: no meconium	Pneumoperitoneum	Dilated small bowel, microcolon with rectosigmoid perforation; primary closure + diversion ileostomy + biopsy; HPE: absent ganglion cells
4 (2 d, preterm, AGA, F)	Abd distension, bilious vomiting, no meconium	Normal	Poor cry & activity	Distended abdomen, visible loops, absent BS, PR: no meconium, wash clear	Question mark sign (contrast enema)	Distended small bowel, microcolon; diversion ileostomy + multiple biopsies; HPE:

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						absent ganglion cells
5 (5 d, term, AGA, F)	Bilious vomiting, passed meconium with stimulation	Normal	Good cry & activity	Mild distension, BS present, PR: meconium staining	Dilated bowel loops	Dilated small bowel, microcolon; diversion ileostomy + multiple biopsies; HPE: absent ganglion cells
6 (1 d, term, AGA, M)	Abd distension, bilious vomiting, no meconium	Polyhydramnios	Poor cry & activity	Distended abdomen, absent BS, guarding & rigidity, PR: no meconium	Pneumoperitoneum	Dilated small bowel, microcolon; diversion ileostomy + perforated site biopsy; HPE: absent ganglion cells

AGA – Appropriate for gestational age; SGA – Small for gestational age; Abd – Abdominal; d – Days; M – Male; F – Female; BS – Bowel sounds; PR – Per rectal; HPE – Histopathological examination.

DISCUSSION

TCA symptoms appear soon after birth as acute intestinal obstruction. As they are rare and hard to diagnose, large multicenter studies are not possible. Single-centre case series are useful for showing local patterns and clinical problems. We present six neonates with confirmed TCA from our centre to show the wide range of presentations and our approach to early diagnosis and management. Common symptoms include difficulty or failure to pass meconium within 48 hours, abdominal distension, and bilious vomiting.[5,6] All our cases showed similar symptom onset in the majority, but also showed significant variability. Two preterm neonates developed spontaneous perforation with pneumoperitoneum; one presented with a meconium ileus-like picture (pellets in the terminal ileum), one had initial meconium passage after stimulation followed by delayed obstruction, and one was associated with Shah-Waardenburg syndrome (white forelock and heterochromia). There is certain variability in neonatal presentation, including classic obstruction, perforation, abnormal meconium transit, and syndromic characteristics. Due to this, TCA can be missed or misdiagnosed initially.[7,8]

Diagnosis of TCA remains challenging. Contrast enema often shows microcolon and a question-mark sign, but a clear transition zone may be absent or located in the small bowel.[1,2] In our series, microcolon was widespread, the question-mark sign was seen in one case, and pellets mimicking meconium ileus appeared in another. Rectal suction biopsy can be inconclusive in TCA, so multiple full-thickness intraoperative biopsies from different colonic levels are essential to map the extent of aganglionosis and confirm preserved ganglion cells at the proximal margin.[1,2] We routinely performed multiple biopsies during diversion ileostomy, which allowed accurate confirmation in all cases.

Surgical care of TCA often begins with diversion ileostomy to decompress the intestine, prevent enterocolitis, stabilise nutrition, and give time for exact level mapping.[1,2] This staged approach is recommended by recent ERNICA consensus and APSA guidelines because primary pull-through in TCA carries high risks of complications.1-3 In our six neonates, initial diversion ileostomy (with perforation repair when needed) was the primary intervention, consistent with current expert recommendations. Children with TCA face higher risks of Hirschsprung-associated enterocolitis, spontaneous perforation, short bowel syndrome after resection, persistent dysmotility, and long-term dependence on parenteral nutrition or stomas.3 Outcomes are generally poorer than in short-segment disease.4 Early diagnosis and diversion can minimise morbidity, as observed in our cases when immediate ileostomy was performed.

CONCLUSION

TCA can present in different ways in newborns, ranging from typical intestinal obstruction to perforation and associated syndromic features. Our six cases highlight the importance of high clinical suspicion, early imaging, and multiple biopsies for early diagnosis. Diversion ileostomy is a safe initial procedure for decompression and diagnosis. Early recognition and staged management can help reduce complications and improve outcomes.

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