

A huge completely isolated duplication cyst complicated by torsion and lined by 3 different mucosal epithelial components in an adult

A case report

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Abstract

Rationale: Intestinal or enteric duplication (ED) does exist as a rare congenital malformation of the gastrointestinal system clinically. It is a separate entity, but can be communicated with the gastrointestinal tract. It is characterized by a well-developed muscular wall and lumen endowed with ectopic mucosa, simulating a portion of normal bowel. A completely isolated duplication cyst (CIDC) refers to an extremely uncommon variant of ED, which is secluded from the alimentary tract and possesses its own exclusive blood supply. Surgical procedure is the treatment of choice, because most often, a definitive diagnosis can only be confirmed intraoperatively.

Patient concerns: A 20-year-old male patient presented with a 10-day history of intermittent episodes of abdominal pain. The pain evolved from dull into progressive and intolerable, accompanied by vomiting, nausea, and abdominal distention.

Diagnoses: Closed-loop small-bowel obstruction with volvulus.

Interventions: The patient underwent an emergency exploratory laparotomy.

Outcomes: A huge CIDC was observed upon operation, which was affixed to the mesentery with only a narrow base, just like a pedicle; 720° counterclockwise twisting around its base was definitely noted, provoking the compromised blood supply. Complete excision of the cyst was performed along its base safely without violating the intestinal tract. Furthermore, the ectopic mucosa of the cyst exhibited 3 different epithelial lining components histopathologically.

Lessons: Clinicians should be aware of the possibility of the existence of a duplication and raise a high index of suspicion in case of equivocal diagnosis, particularly in adult population. A low threshold for surgical management should be recommended in order to prevent lethal outcomes.

Abbreviations: CIDC = completely isolated duplication cyst, CT = computed tomography, ED = enteric duplication, EDC = enteric duplication cyst, US = ultrasonography.

Keywords: completely isolated duplication cyst, intestinal or enteric duplication, mucosal lining, torsion

1. Introduction

As a potentially life-threatening disease, an enteric duplication (ED) or intestinal or enteric duplication cyst (EDC) has all the

time been perplexing pediatric surgeons. It is really a challenging task to make a clinical diagnosis mainly due to its rarity and nonspecific presentations, unless complications ensue unexpectedly. In addition, it affords little opportunity to elucidate its mysterious nature and characteristics.

An EDC is an unusual congenital deformity of the alimentary system, which is a separate entity invested with a cystic appearance, but at the same time is in intimate contact or communication with the alimentary tract.^[1] This entity consists of a wide variety of cystic lesions originating from different sites alongside the digestive tract between the mouth and the anus in conjunction with their neighboring organs, with a predilection for the small bowel, especially the ileum.^[2,3] It takes place most commonly in infants and children, with an estimated incidence of approximately 1 in 4500 births,^[4] but is exceedingly uncommon in adults.

A completely isolated duplication cyst (CIDC) refers to an extremely rare variant of EDC, which does not contact with or is secluded from the alimentary tract and possesses its own exclusive blood supply.^[5] A CIDC occurs much more scarcely, with a prevalence of about 1 in 10,000 live births,^[6] and to the best of our knowledge, there have been only 10 cases of CIDC in adults ever published in English literature,^[5,7–15] among which 2 experienced malignant degeneration.^[12,14] Herein, we reported a case of a 20-year-old male patient who presented with a huge CIDC, which

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Informed written consent has been obtained from the patient for the publication of this case report.

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served as the lead point for the cystic torsion, adding up to the increased difficulty for diagnosis and rendering the mandatory emergency laparotomy. Interestingly, the ectopic mucosa of the cyst was lined with 3 different epithelial components histopathologically. To date, such a case has never been reported yet.

2. Case report

A 20-year-old male patient was submitted to the emergency with a 10-day history of intermittent episodes of abdominal pain without any obvious predisposing cause. The pain seemed dull at first, but gained progressive and intolerable gradually, which was then accompanied by vomiting, nausea, along with abdominal distention. The discomfort had not settled with conservative treatment. The patient was otherwise healthy without such an onset ever before.

On examination, he appeared anguished, but still maintained normal vital signs. His abdominal examination revealed tenderness over the lower abdomen, with no rigidity, rebound tenderness, or voluntary guarding. No palpable mass was palpated and the bowel sound was as usual. The laboratory investigations were only remarkable for an elevated white blood cell count of $17.28 \times 10^9/L$, with NEUT% 85.5%. The plain abdominal X-rays provided no special findings. The ultrasonography (US) visualized significantly dilated bowel loops together with a small amount of abdominal fluid. The contrast-enhanced computed tomography (CT) scan (Fig. 1) was subsequently performed and demonstrated multiple evidently dilated cystic-like small-bowel loops with thickened walls, communicating with each other. There was twisting of mesenteric vessels around each other suggesting the whirlpool sign, apart from a mild portion of ascites. Taken together, features on the CT scan were highly indicative of closed-loop small-bowel obstruction with volvulus to be the likely cause. Therefore, the patient was admitted to the department of general surgery immediately. After his endorsement of the informed consent, he underwent an emergency exploratory laparotomy.

At surgery, the intra-abdominal exploration demonstrated that a huge cystic-like object was located in the ileal mesentery 45 cm

proximal to the ileocecal junction and assumed a smooth surface. It was apparently distended and ischemic but without obvious gangrene or necrosis. On first impressions, this cyst appeared to be a swollen small bowel loop (Fig. 2). Whereas, it was neither connected nor adhesive to the adjacent bowel. In fact, this object was only affixed to the mesentery with a narrow base, just like a pedicle constricting its feeding vessel, and it was in no small part mobile, blind and secluded from the bowel lumen. Furthermore, 720° counterclockwise twisting around its base was definitely noted, which provoked the compromised blood supply. There was approximately 50 mL turbidly purulent ascites additionally. Subsequently, following a clockwise untwisting, the cyst was excised along its base safely without violating the intestinal tract.

Grossly, the resected specimen measured 70 cm in length and 8 cm in maximal diameter, which was unilocular and contained massive hemorrhagic fluid intermixed with clots. Histopathology (Fig. 3) illustrated that the cystic wall was uniquely composed of a heterotopic mucosa combined with intact underlying well-defined smooth muscle layers, resembling a part of normal bowel tract. All these findings confirmed a final diagnosis of an ED consistent with congenital malformation. A variety of mucosal lining types were displayed throughout the lumen: segmented squamous epithelium, focal gastric glandular mucosa of foveolar and oxyntic type, in addition to patchy intestinal metaplasia. The postoperative recovery course was uneventful without any complications or events. The patient was discharged on the 7th postoperative day and still remained well over the routine follow-up.

3. Discussion

An ED is quite infrequently seen in an adult. Reportedly, it usually occurs during the neonatal or childhood period, mostly before the age of 2,^[16] and is slightly predominant in males.^[16] Although this extraordinary anomaly is an independent entity, most often it is adherent to or contiguous with the main intestinal tract. However, it is constantly variable in location and anatomy. It can be sited on either the mesenteric or the antimesenteric side

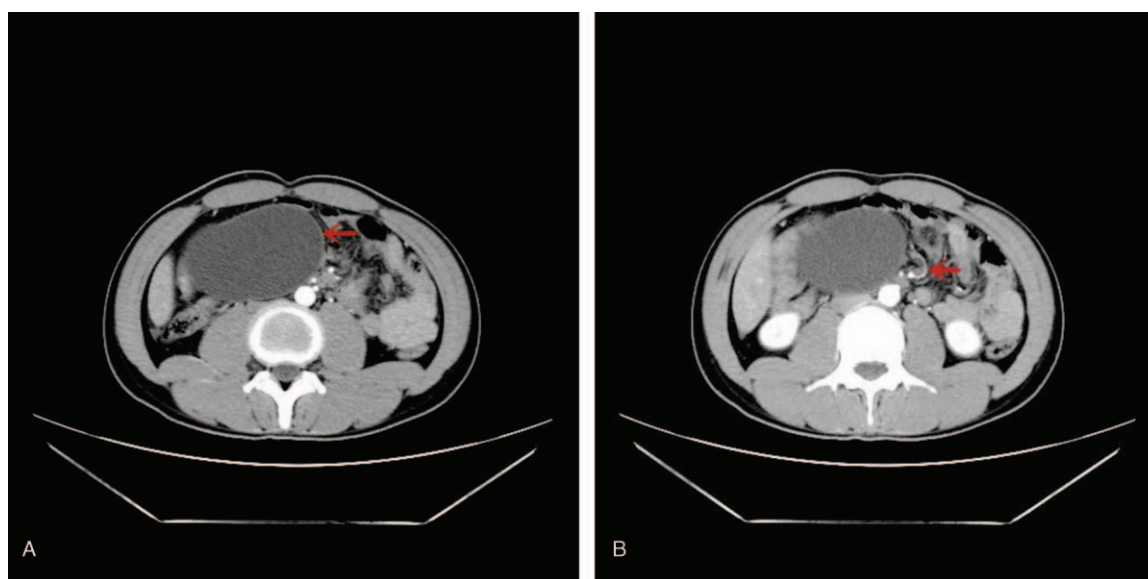


Figure 1. Features of the duplication on computed tomography scan. (A) A demonstration of a well-margined cyst mistaken initially as a distended bowel loop (arrow), which was definitely an intestinal duplication verified intraoperatively and retrospectively, full of hemorrhagic fluid. (B) A description of twisting of mesenteric vessels around each other at the base of the cyst (arrow), suggestive of the whirlpool sign.

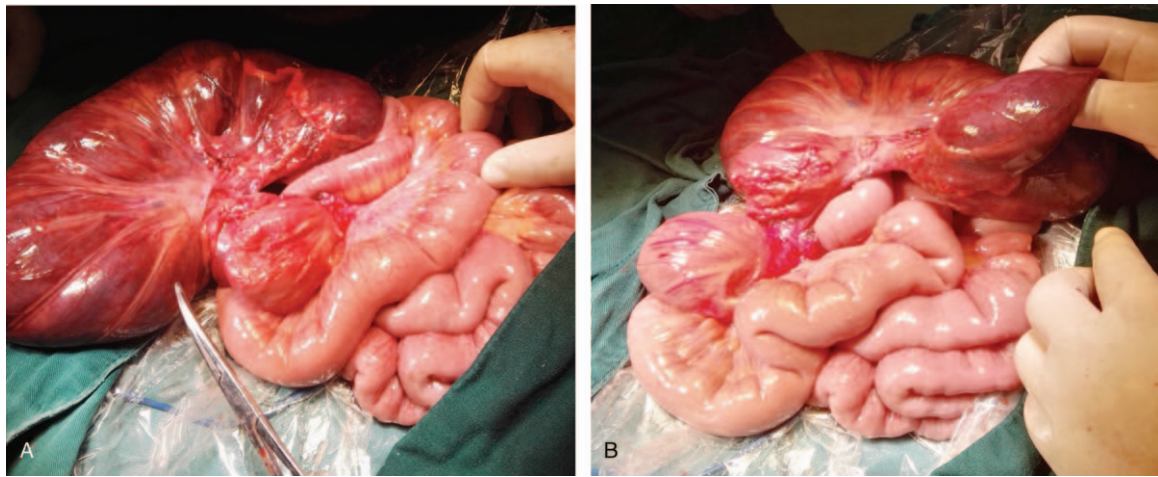


Figure 2. Intraoperative findings of the detorsioned duplication cyst. (A) The distended, ischemic, and separate cyst, measuring 70 cm in length and 8 cm in maximal diameter; (B) the attachment of the cyst to the ileal mesentery with a pedicle, without any communication with bowel.

of the bowel wall, with a mesenteric preponderance,^[1] and its length ranges from few centimeters to whole length of the small intestine.^[17] Most of the time, it is single, while concurrent multiple cysts are exposed in 10% to 15% of cases. It appears unilocular mostly, but sometimes could be multilocular, or even

multilobulated.^[18] Frequently, a duplication is mobile, and can be observed as a palpable mass in 50% of cases.^[19] Rarely can an ED have its own autonomous blood supply independent from the neighbouring bowel tract; conversely, it shares the common vessel system with the mesentery.^[20] Even some additional

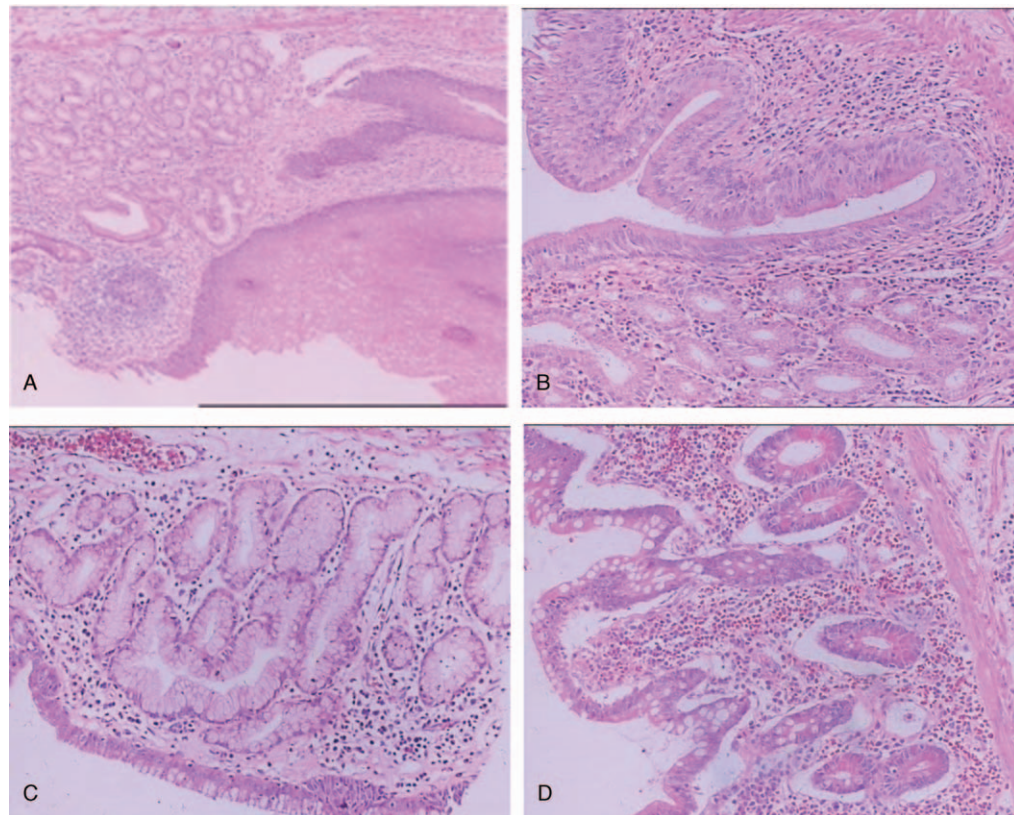


Figure 3. Histopathological configuration of the duplication. (A) The cystic wall containing an ectopic mucosa with a variety of epithelia linings covering the cystic cavity, and overlying the smooth muscle layer (hematoxylin and eosin [HE], $\times 40$). (B) An illustration of the focal area of mucosal transition between squamous and glandular epithelia (HE, $\times 100$). (C) A depiction of the small portion of gastric type mucosa with foveolar and oxyntic glands easily appreciated, secreting mucous substances (HE, $\times 100$). (D) A representative section of formation of the intestinal metaplasia, showing a sheet of enteric type mucosa rich in goblet cells and Paneth cells, which were infiltrated with myriads of erythrocytes (HE, $\times 100$).

associated anomalies have been encountered in 50% of patients, the majority being vertebral defects.^[1,16] An ED can also be correlated with malrotation.^[21] Our current patient was not found to have other anomalies or malrotation, with the duplication located in the mesentery separately.

By definition, an ED or EDC is characterized by a well-developed muscular wall along with a lumen lined by ectopic mucosa,^[11] which bears a striking resemblance to a portion of the normal bowel tract. It is endowed with the similar layered muscles with the normal bowel wall, and frequently even shares a common muscular wall. The ectopic mucosa is typically lined by gastrointestinal-type epithelium ranging from gastric to rectal component,^[22] which is usually compatible with the adjacent bowel, while different epithelial constituent other than gastrointestinal origin has been sometimes identified, commonly being respiratory epithelium.^[22] Normally, a single epithelial lining is present, the majority being gastric,^[2] although a combination of multiple types of epithelia coexisting in the same duplication has occasionally been disclosed.^[1,23] Besides, infrequently, tissues like an annular or aberrant pancreas are also observed,^[24] but cartilage is never shown within the wall.^[25] In our case, squamous epithelium, which is of disparate origin and seldom emerges in the normal abdominal cavity, in combination with 2 other gastrointestinal-type epithelial compartments, uniquely comprised the mucosal lining.

Functional secretion of the ectopic mucosa of an ED tends to produce a clear mucoid fluid in the hollow lumen,^[20] leading to distention of the cyst. In terms of ectopic gastric mucosa, excessive acid secretion may give rise to ulceration, erosion, bleeding, or perforation.^[26] By contrast, heterotopic pancreatic tissue could trigger increased pancreatic enzymes contained in the fluid,^[27] even hypoglycemia.^[26] An ED uncovered in the neonates is benign, but can develop malignant transformation due to the persistent presence and stimulation of ectopic mucosa, which is solely seen in adults^[4,11] and in the form of adenocarcinoma.^[28] There was no evidence of dysplasia or malignancy in our case, and the mucosal secretion potentially contributed to its presentations and complications.

Generally speaking, there are 2 chief categories of duplication according to morphology^[20,29]: one is tubular, while the other cystic or spherical. The tubular communicates with the bowel lumen, whereas the cystic representing the vast majority of cases, does not. The duplication in our case was reasonably cystic, with no communication. Meanwhile, on the basis of the vascular pattern, Li et al^[30] have classified small EDs as type 1 or parallel type (74.6%) and type 2 or intramesenteric type (24.4%). The duplication in our case belonged to type 1 logically: the cyst resided on one of the leaves of the mesentery and its feeding artery was separate from that of the bowel. The most important implication of this classification may possibly lie in its determination of complete excision without disturbing the continuity of the bowel, just as depicted in our patient.

A CIDC was present in our patient, which had a dedicated vascular peduncle, in keeping with description in some other literature.^[11] It just hung free on the mesentery, which made it susceptible to twisting around the peduncle, serving as the lead point for torsion. Plausibly, there has already raised a speculation that an ED might predispose the patient to volvulus.^[31]

As to the pathogenesis, it is probably related to embryonic development disorders. Several major theories have been hypothesized,^[29] to explain the development of duplication arising in different sites, such as vacuolization, caudal twinning, diverticularization, as well as split notochord theories and so

on.^[32] Nevertheless, none of them have been persuasive enough alone. Thence, the precise mechanism still remains poorly understood.

Regarding its symptoms it is particularly difficult to distinguish an ED from other intra-abdominal cystic etiologies.^[8,19] Besides, it is quite confused with a Meckel's diverticulum when locating on the antimesenteric side. Its manifestations may be vague, and lack of specificity, mimicking other diseases.^[3,33] Although it can present with acute abdomen during the neonatal or childhood period, even progressing to unexpected death, it could also remain asymptomatic until at school age or adulthood.^[3] Nonetheless, the vast majority of cases would develop symptoms in the first year of life.^[34] Of importance, most often, only can enough attention be paid to the very cases due to inadvertent manifestation of complications, including intestinal obstruction, perforation or cystic rupture, volvulus or cystic torsion, infection, or malignancy.^[4]

Numerous imaging modalities may help in diagnosis of an ED. US plays a crucial role in the evaluation of a duplication, and has the capability of showing the characteristic double wall signs or gut signature signs.^[29] Many thanks to its improved accuracy, prenatal US has identified a good number of cases of duplication in utero.^[1] However, this very sign could sometimes be misleading,^[34] which is also observed in cysts of other geneses (false positive),^[35] such as mesenteric cyst, Meckel's diverticulum, ovarian cysts, and cystic teratoma, and so on. Thus far, only 20% to 30% of lesions with prenatal US have been detected,^[4] and an overall sensitivity of merely 55% has been obtained pertaining to the diagnosis of ED actually.^[36] Magnetic resonance imaging, which is recommended to prenatal utility as well, is as diagnostic as US, but unable to provide any more useful information.^[20] Barium studies may be helpful if the duplication is tubular,^[1] communicating with the bowel lumen, and a gastrointestinal endoscopy might be equally feasible upon such an occasion. In the context of gastrointestinal bleeding, Tc-99m pertechnetate radionuclide scan is liable to discern the heterotopic gastric mucosa, which aids in the discrimination of Meckel's diverticulum from a duplication when locating on the antimesenteric border of the small bowel, with an up to 75% sensitivity.^[37] Nonetheless, it is worth noting that heterotopic gastric mucosa does emerge in only 20% to 30% of duplication cysts,^[38] which dramatically limits the detectable usefulness of isotope studies.^[39] Enhanced CT scan is a more common and desirable imaging tool, offering further characteristics of the lesion along with delineation of the surrounding structures,^[33] which most likely indicates the necessity of immediate management.

In our patient, US failed to reflect the duplication, with merely a perception of puffy bowel loops, which was principally ascribed to lack of suspicion, little familiarization with its appearance, as well as obstacles created by trapped gas in the enlarged bowel loops. But we are convinced that such US findings of dilated bowel loops would be highly suspicious of an ED or intra-abdominal cyst should it take place in a neonate or child. In light of the noteworthy information of CT scan indicative of bowel obstruction and volvulus, we determined to take prompt measures for surgical treatment, thus potentiating the final diagnosis and satisfactory prognosis. Nevertheless, on review of the CT scan and consultation with the radiologists, we are confident that the markedly enlarged bowel loop (Fig. 1A) was definitely an ED, in accordance with the patterns described in the literature.^[29,34]

Unfortunately, a definitive diagnosis in most cases is only confirmed incidentally, especially intraoperatively,^[1] irrespective

of the assist of imaging methods, just as shown in our case. Once torsion or volvulus occurs, it can progress to strangulation or necrosis abruptly, thus contributing to the deteriorated risk of morbidity and mortality. Hence, a low threshold for surgical management should be recommended in cases of suspected diagnosis in adults, so as to avoid lethal aftermaths.

The most preferred management is total excision of the lesion without bowel wall compromise, which is exclusively suitable to an isolated duplication like CIDC, just as described in our patient. Otherwise, resection of the duplication along with a segment of the adjacent bowel is mandatory on account of the intimate attachment and involvement. Nonetheless, long segment bowel resection should be discouraged as far as possible, in avoidance of short bowel syndrome, and upon such a setting, some alternative surgical options, such as staged approaches or mucosal stripping, would be more beneficial as appropriate.

4. Conclusion

An ED does exist as a rare form of congenital defect of the gastrointestinal system clinically. Given its atypical presentations and the challenge to make a diagnosis, clinicians should be aware of the possibility of its existence and raise a high index of suspicion in case of equivocal diagnosis, particularly in adult population. Once taking the suspicion into consideration, just as depicted in our patient, either an exploratory laparotomy or, whenever possible, laparoscopy should be designated in order to verify the diagnosis, to facilitate a timely treatment as well as to eliminate progression of severe complications.

Author contributions

Ai Xiao-Ming, Lu Jin-Jing, and Ho Li-Chen conceived the research idea, designed this study, as well as drafted the manuscript. Ai Xiao-Ming, Han Lu-Lu, and Yue Xiong completed the clinical data collection and performed histological examination. Yang Nian-Yin and Zhang Hong-Hai interpreted the clinical data and revised the manuscript. All the authors read and approved the final manuscript.

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