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Completely Isolated Enteric Duplication Cyst and Incidental Neuroendocrine Tumor of the Appendix: A Case Report

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Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
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Funds Collection G

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Conflict of interest: None declared

Patient: Female, 26-year-old
Final Diagnosis: Completely isolated enteric duplication cyst and appendiceal neuroendocrine tumor
Symptoms: Dysmenorrhea
Medication: —
Clinical Procedure: —
Specialty: Oncology • Surgery

Objective: Rare disease

Background: Completely isolated enteric duplication cysts (CIDCs) are rare malformations that can occur at any site in the gastrointestinal system. This report describes a woman with a CIDC and an incidental appendiceal neuroendocrine tumor (ANET).

Case Report: A 26-year-old woman who presented with dysmenorrhea was assessed by ultrasound (US), which revealed a pelvic mass. Other imaging modalities, including magnetic resonance imaging (MRI), failed to clarify the origin of the mass. Intraoperative findings during diagnostic laparoscopy revealed an isolated, ovaloid mass with autonomous peristalsis and a short pedicle towards the root of the ileal mesentery. In addition, the appendix appeared enlarged with a hardened consistency. The mass was resected and an appendectomy performed laparoscopically. The pelvic mass was diagnosed as a CIDC and the appendix was incidentally found to contain a pT3Nx carcinoid tumor. Based on histological examination and guidelines of the European Neuroendocrine Tumor Network (ENET), the patient later underwent a laparoscopic right hemicolectomy.

Conclusions: CIDC in adulthood is very rare, especially when combined with an incidentally discovered pT3Nx appendiceal carcinoid tumor. Neither US nor MRI was able to provide a precise preoperative diagnosis. Diagnostic laparoscopy clarified the nature of the mass and revealed a lesion missed during the preoperative workup. Because of the diagnosis of ANET, the patient subsequently underwent a laparoscopic right hemicolectomy.

MeSH Keywords: Appendiceal Neoplasms • Carcinoma, Neuroendocrine • Laparoscopy • Mesenteric Cyst

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Background

Enteric duplication cysts (EDC) are uncommon congenital malformations, which can arise anywhere in the gastrointestinal system, usually on the mesenteric side, occurring in an estimated 1 of 4000–5000 births [1–6]. EDCs occur more frequently in the small bowel (jejunum and ileum, 47%) than in the colon (20%), esophagus (17%), stomach (8%) and duodenum (2%). These cysts are characterized by a round or tubular shape and maintain an intimate communication with the normal alimentary tract, usually sharing the same muscular coat and blood pedicle [5].

These features are lacking in completely isolated duplication cysts (CIDC), a rarer entity with an incidence of 1 in 10,000 births. CIDCs have their own vascular supply and have no communication with the bowel [3,5,7–9]. CIDCs are normally detected in newborns and infants, but rarely in adults. This report describes a young woman who visited our institution due to menstrual irregularities and was later diagnosed with CIDC and an incidental appendiceal neuroendocrine tumor.

Case Report

A 26-year-old woman was referred for surgery in March 2019 after a year of oligo- and dysmenorrhea, without any other symptoms. The patient was a non-smoker, had no comorbidities and had never been pregnant. A transvaginal and pelvic ultrasound (US) scan in April 2018 had revealed a thin-walled, avascular mass in the right iliac fossa, medial to the right ovary with no mutual communication (Figure 1). Abdominal magnetic resonance imaging (MRI) in May 2018 revealed a 64×44×54 mm cystic lesion filled with fluid in the right para-adnexal space, which was diagnosed as a sactosalpinx. Both ovaries were considered normal in morphology and dimension (Figure 2). A second transvaginal US scan in October 2018 confirmed these findings, as well as showing a synchronous peristaltic activity with the intestinal loops. Differential diagnosis suggested a diverticulum or a mesenteric cyst. An abdominal US scan with oral contrast in December 2018 did not highlight any enhancement inside the mass, indicating a hyperechoic sediment. The mass was considered a cystic lesion that derived from the ovary. A transvaginal US scan in February 2019 revealed a major increase in the dimensions of the mass, which was 9 cm in diameter. Peristalsis was again observed.

Because of the increase in the dimensions of the mass and the lack of a definitive diagnosis, the patient was referred to our Minimally Invasive Surgery Unit. where she was scheduled for a diagnostic laparoscopy with eventual mass excision. She underwent an elective diagnostic laparoscopy on March 29, 2019. Intraoperative examination showed that her uterus, ovaries, Fallopian tubes and large ligament were normal. Meticulous exploration of the intestinal loops, however, revealed an isolated, ovaloid mass with autonomous peristalsis, diffuse superficial vascularization, and a short pedicle at the root of the ileal mesentery (Figure 3). Her appendix appeared enlarged with a slightly hardened consistency, suggesting an inflammatory process but with no evidence of malignancy. The mass in her intestinal loops and her appendix were removed laparoscopically. Her postoperative course was uneventful. She was able to tolerate a semiliquid diet on the first postoperative day and was discharged the following day.

Histological examination of the intestinal mass showed a cystic layer covered by smooth muscle tissue and an inner mucosa of bowel-like pseudo-stratified ciliate epithelium with some

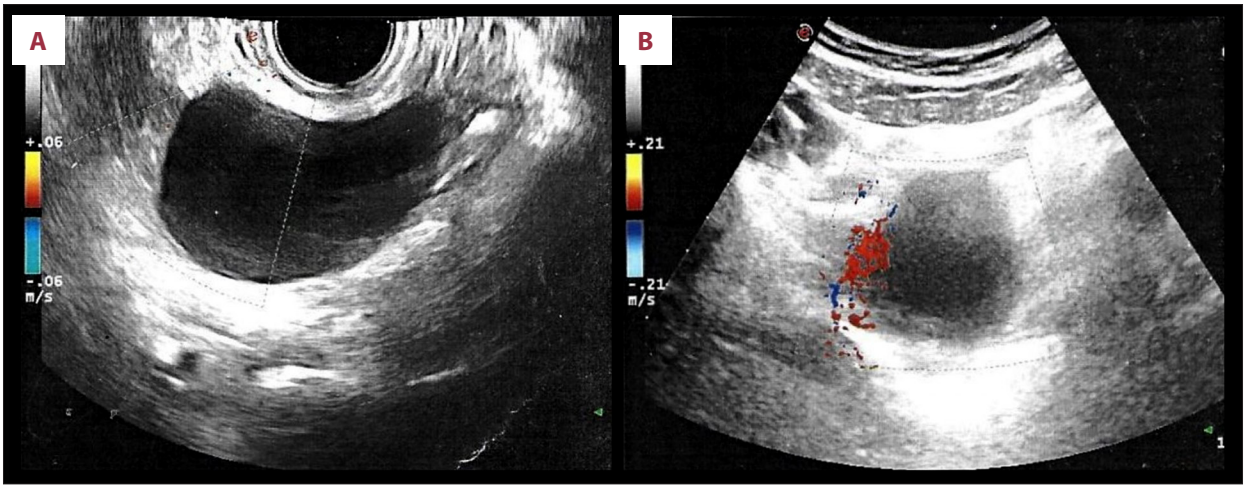

Figure 1. (**A**) Endocavitary ultrasound (US) (Philips Model No: iU22, Bothell, WA), showing an anechoic rounded mass with well-defined margins and measuring 64×44×54 mm, next to the right fallopian tube. (**B**) Color-Doppler module did not show any intralesional vascular signs.

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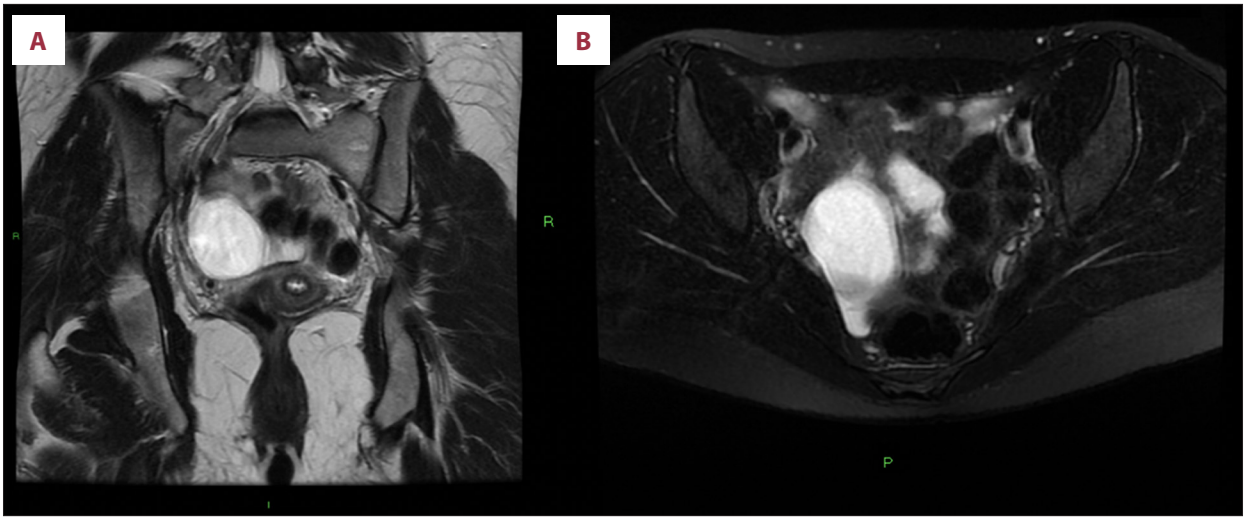

Figure 2. MRI of the pelvis using a 1.5-T MRI scanner and Sense Torso Coil. (**A**) TSE T2 sequence (coronal plane). (**B**) TSE STIR (axial plane) of the lesion, showing low signal intensity on T1-weighted sequences and high signal intensity on T2-weighted sequences. The MRI features of the mass in the right para-adnexal space, which suggested the presence of a cystic lesion, led to a suspected diagnosis of sactosalpinx.

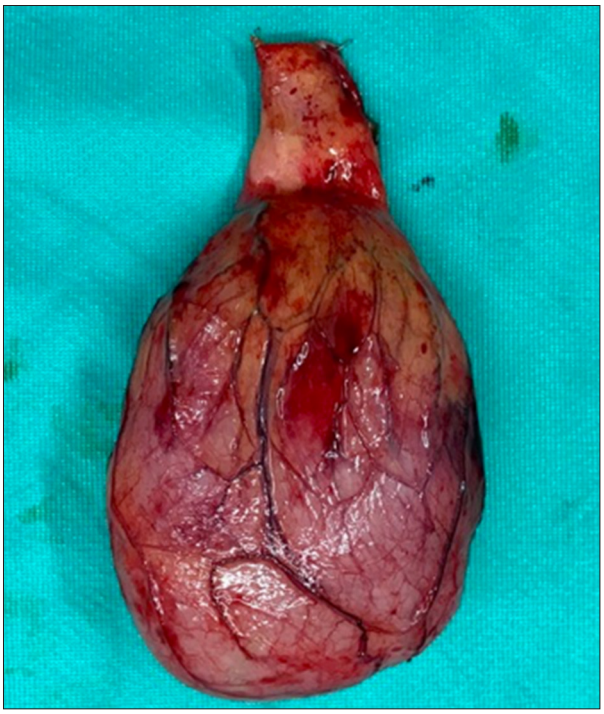

Figure 3. Macroscopic appearance of the resected CIDC.

milky-ochreous material inside (Figure 4), a finding consistent with a diagnosis of CIDC. The appendix contained a 4.5-cm long yellowish neoformation obstructing the lumen and invading the entire appendiceal wall and mesoappendix. This lesion was found to be an incidental neuroendocrine tumor (NET, G2 appendiceal carcinoid) with involved surgical margins, positive for both synaptophysin and chromogranin A, and with a 2–3% Ki67. The tumor was classified as a pT3Nx appendiceal neuroendocrine tumor (ANET) according to the 2010 criteria of the World Health Organization (Figure 5).

A positron emission tomography/computed tomography (PET/CT) scan for NET tumor staging in May 2019 showed no evidence of pathological enhancement. Because the CIDC lacked malignant features, there was no need for further diagnostic examinations, including assessments of tumor markers. After multidisciplinary discussions and based on the guidelines of the European Neuroendocrine Tumor Network (ENET), the patient underwent a laparoscopic right hemicolectomy on July 5, 2019. Histological examination revealed the complete absence of tumor tissue in the right colon and in the 32 lymph nodes that had been removed. The patient's post-operative course was uneventful, and she was discharged on postoperative day 5.

Postoperatively the patient continues to experience irregular menses. Her gynecologist did not indicate further treatment but rather regular follow-up. She is now well and has been referred for oncologic follow-up, which established that she required no further treatment.

Discussion

The natures of EDCs and CIDCs remain uncertain, although they have been linked to embryonic developmental disorders. Several hypotheses have attempted to explain their etiology [2,3,10,11]. These include 1) aberrant luminal recanalization, in which a CIDC is derived from an aberrancy in the canalization process, resulting in a parallel tract; 2) abortive twinning; 3) a split notochord; 4) persistent embryologic

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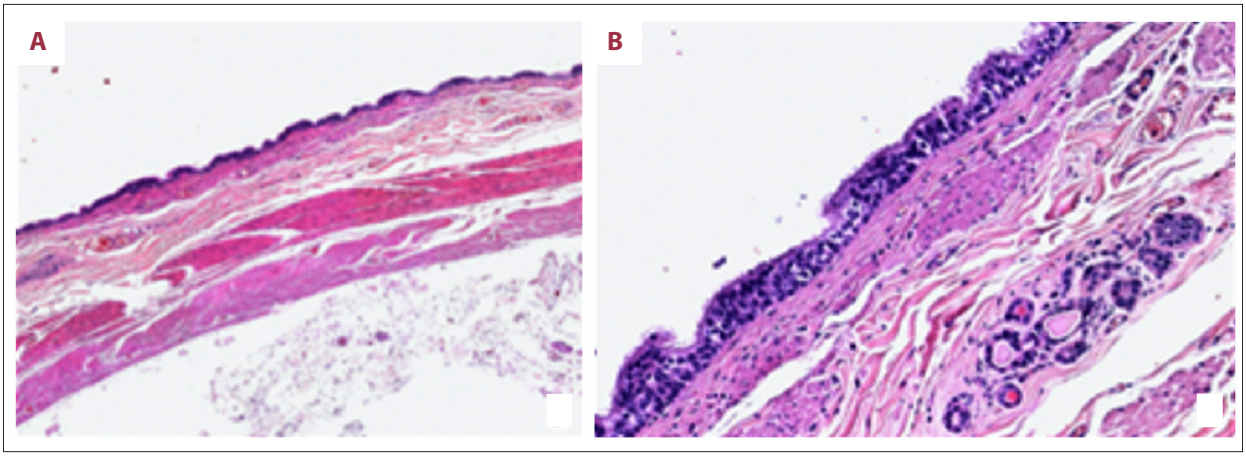

Figure 4. Histologic examination of the CIDC, showing (**A**) a duplicate wall composed of a mucosa, submucosa and a muscolaris propria with two muscular layers (H&E, 20×) and (**B**) lining epithelium made up of columnar pseudostratified ciliated cells with glands in the submucosa (H&E, 100×).

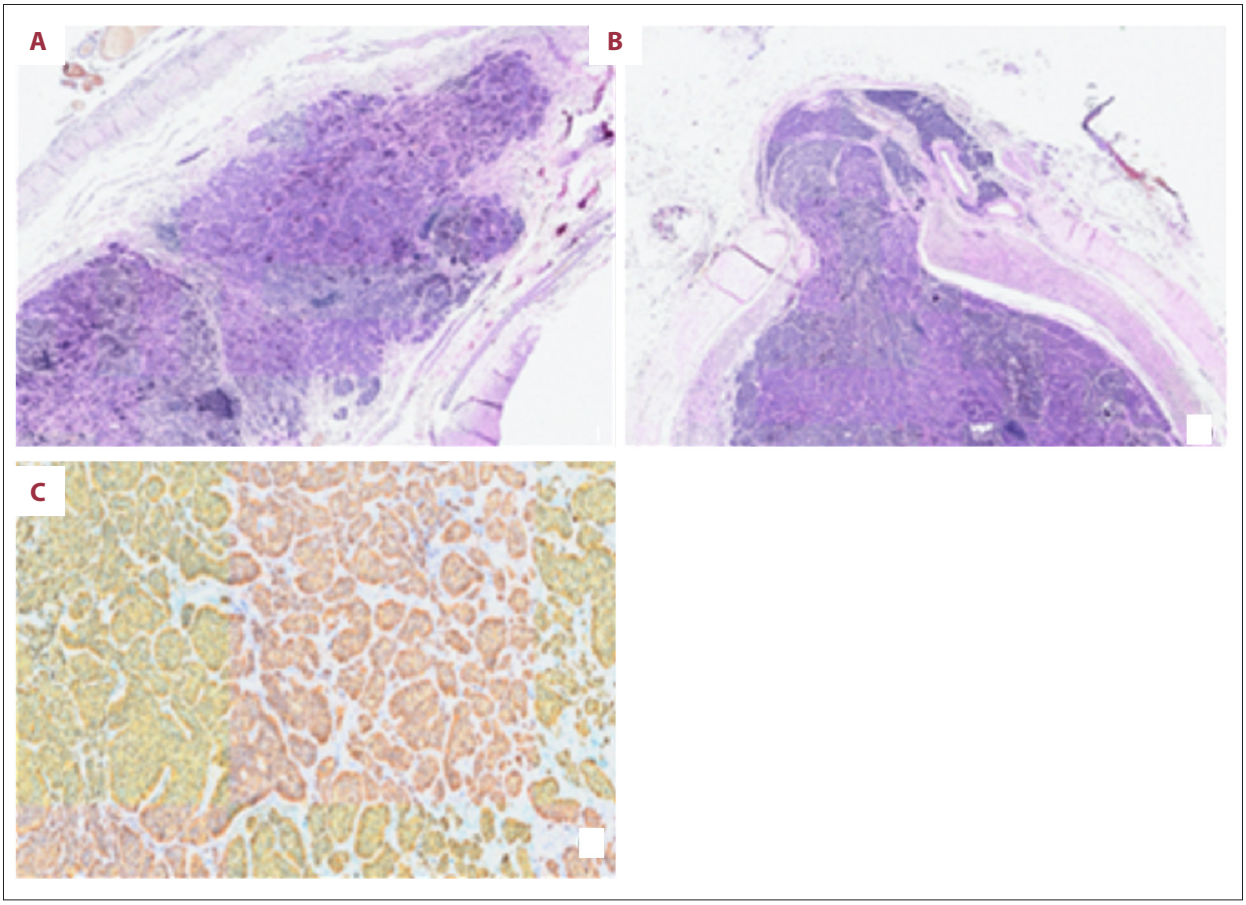

Figure 5. Histologic examination of the appendix, showing (**A**) the tip of the appendix with nests of cells containing little cytoplasm and salt and pepper nuclei; (**B**) carcinoid infiltration of the appendiceal wall and periappendicular tissue; and (**C**) acidophilic granules in the cytoplasm (each H&E, 20×). Insert: Cromogranin A positivity.

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diverticula; 5) sequestration of the fetal gut; 6) vacuolization; and 7) diverticulum torsion or a vascular accident resulting in its detachment from the intestinal wall [12].

Most EDCs are detected in early childhood, although they can also be found incidentally in adults. CIDCs are detected primarily in newborns or during early childhood. CIDCs are frequently asymptomatic, or they may show generic symptoms, such as non-specific abdominal pain, nausea and vomiting. Other symptoms can include obstruction, intussusception, jaundice, pancreatitis and gastrointestinal hemorrhage [10,13]. CIDCs are linked to other malformations, such as biliary or intestinal atresia, malrotation, imperforate anus, double gallbladder, double uterus, partial gastric diverticulum, complete large bowel duplication, situs inversus and intraspinal neuroenteric cyst [10,13]. CIDCs in adults are extremely rare, with fewer than 50 such patients reported to date, and are usually diagnosed incidentally [3,4,6]. Despite our patient undergoing regular prenatal screening, CIDC was not detected, precluding a determination of mass development.

CIDCs may occasionally present in acute settings as palpable and painful masses or as acute hemorrhage resulting from ulceration [1,2]. Malignant transformation within the duplication has been reported, although it is very rare. Lesion resection is therefore required to prevent complications, allow a precise histopathologic diagnosis, and rule out a possible malignancy [9,14–16].

CIDCs are hollow structures with a composition and histomorphology similar to those of the nearby bowel, as well as being autonomous with no luminal connection. The serosa, muscular layer and submucosa of CIDCs are apparently similar to those of the nearby bowel, but the mucosa layer is usually different. The epithelium of a CIDC may be similar to the nearby tract [17] or heterotopic (gastric, pancreatic and respiratory). A different type of epithelium can alter the clinical presentation, as acid secretion may lead to the development ulceration, erosion, and bleeding up to perforation [18]. Enzymes in pancreatic tissue may be activated, resulting in a pancreatitis-like syndrome [19]. CIDCs are therefore classified based on the adjacent portion of the alimentary tract or mucosal layer.

Because CIDCs have non-specific symptoms, it is often difficult to distinguish them from other intra-abdominal cysts [2,17,20], such as Meckel's diverticulum (especially when the cyst is located on the antimesenteric side), ovarian cyst, cystic teratoma, ganglioneuroma, and mesenteric cyst [1,2,4,5,10]. Because US was unable to precisely define the nature of the mass, MRI was performed, as it has been found more valuable than CT in assessing abdominal fluid masses [1,21–23]. Moreover, MRI avoids exposing a young fertile female patient to the radiation associated with CT [1,21–23]. Nevertheless, the only pathognomonic sign suggestive of CIDC was the peristaltic movement observed on US.

The uniqueness of this case was due to the concomitant presence of an incidental appendiceal NET (G2). Pre-operative US and MRI images, even after retrospective review, showed no indication of a pathological appendicular mass. Nevertheless, the appendix was intraoperatively removed because of its appearance. Appendiceal NETs constitute 30–80% of all appendiceal tumors [24], making them the most frequent type of NET. Most patients with these neoplasms have an excellent prognosis, with a 5-year survival rates close to 100% in patients with lower staged tumors [25]. Appendiceal NETs are usually detected intraoperatively or on histological examination after appendectomy, as they do not normally have a tumor-related symptomatology [24]. Current WHO guidelines classify this patient as a having a T3G2Nx appendiceal neuroendocrine neoplasm (ANEN), since it had departed from the appendiceal base to invade the mesoappendix. Because this ANEN had negative prognostic features (G2, size >2 cm, mesoappendiceal extension) and based on ENETS guidelines, this patient later underwent a right hemicolectomy.

To our knowledge, only one previous study described a similar patient with a concomitant double enteric duplication and an ANET in an emergency setting involving an acute abdomen. Despite differences in their clinical presentations, their pre-operative work-up was similarly challenging, as radiological imaging was not diagnostic, with the lesion identified only after diagnostic laparoscopy [26].

Conclusions

CIDCs are extremely rare, vary in clinical presentation, and are more often found during childhood than in adults. The case reported in this study is even rarer, due to the adult age of the patient and the synchronous presence of an appendiceal NET. To our knowledge, this is the second patient who has presented with an isolated enteric cyst and a neuroendocrine neoplasm of the appendix.

Preoperative diagnostic work-up was particularly demanding, as both US and MRI scans could not determine a definitive diagnosis. Laparoscopy, however, was excellent in diagnosing a missed pathology and performing a simultaneous resection, resulting in rapid recovery and a shorter hospital stay.

Malignant transformations can occur within enteric duplications, with surgical removal preventing this occurrence.

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