

Thoraco-abdominal enteric duplication cyst in association with neurenteric cyst, axial skeletal anomalies, and malrotation

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We report a case of a 2-year old boy with cervicothoracic deformity with vertebral rib anomalies, neurenteric cyst, separate thoracoabdominal enteric duplication cyst, concurrent intestinal malrotation, and dextroposition of the heart. This combination of abnormalities is very rare. When these lesions are suspected, the patient must be investigated carefully. This case is presented to show the importance of cross-sectional imaging (MR and CT) for surgical planning.

Introduction

The split notochord syndrome was first described by Bentley and Smith in 1960, and includes a spectrum of abnormalities involving the vertebral column, alimentary tract, and central nervous system (1). The notochord and endoderm are near neighbors during the development of the embryo, and therefore several authors have postulated that a duplicated or cleft notochord might lead to an adhesion of the endoderm to the ectoderm, resulting in the spectrum of abnormalities described as the split notochord syndrome (1, 2).

Only one other report of similar combination of abnormalities has been published, but that case featured a meningocele instead of a neurenteric cyst, with the addition of hypoplastic left upper extremity and camptodactyly

(3). Another report of transdiaphragmatic duodenal duplication in combination with neurenteric cyst has appeared in the literature (4).

Case report

A 2-year old boy presented with nonbilious emesis and abdominal pain. Past medical history included full-term pregnancy and a history of dextrocardia. A plain radiograph of the chest demonstrated displacement of the mediastinum into the right chest by mass effect in left chest (Fig. 1). Low-dose CT (CTDIvol) of the head showed no evidence of intracranial abnormality. Low-dose CT(CTDIvol) of the chest and abdomen demonstrated multiple congenital fusion and segmentation vertebral anomalies from C5 to T4. The left first four ribs were absent. Additionally, an anterolateral thoracic probable myelocele at the level of T4 was noted. In the left posterior chest were septated cystic structures extending down toward the aortic hiatus at the diaphragm. These cystic structures appeared separate from but contiguous to the myelocele, as proven by intraspinal contrast placed proximally (Fig. 2). Dextroposition of the heart into the right chest with maintenance of normal chamber relationships was noted. The dextroposition appeared to be secondary to mass effect of the cystic structures in the left hemithorax, associated with underdevelopment of the left lung. Low-dose CT (CTDIvol) of the abdomen showed continuation of the cystic structure through the diaphragm from the left hemithorax crossing into the right upper abdomen just anterior to the aorta between the celiac axis and superior

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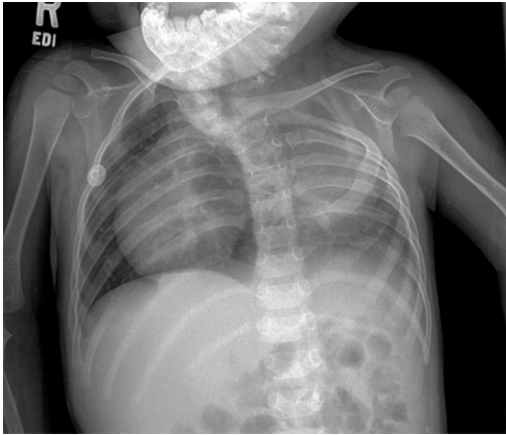


Figure 1. 2-year-old male with thoraco-abdominal enteric duplication cyst. AP radiograph of chest demonstrates mass effect in left hemithorax, displacing the mediastinum to the right, and cervico-thoracic vertebral and rib anomalies on the left side.

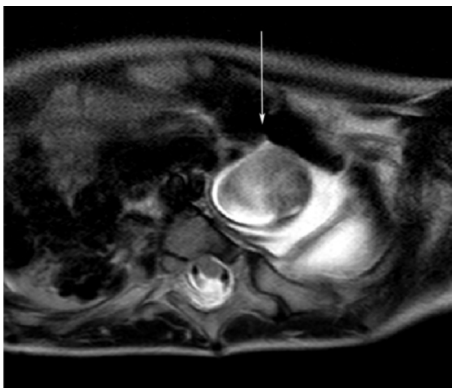


Figure 2. A. 2-year-old male with thoraco-abdominal enteric duplication cyst. Axial T2W MRI at the level of the upper thoracic cord. A left lateral myelomeningocele sac is positioned anterior to a fluid-enteric cyst. This sac shows loss of T2 signal secondary to turbulent CSF flow (arrows). B. Sagittal T2W MRI of the spine shows the same myelomeningocele positioned ventral to the spinal canal and superior to the fluid-filled enteric cyst (arrow).



Figure 3. 2-year-old male with thoraco-abdominal enteric duplication cyst. Coronal (A) and sagittal (B) reformats demonstrate myelographic contrast in neurenteric cyst (arrow) adjacent to thoracic portions of duplication (d) in posterior mediastinum extending across diaphragm into upper and lower abdomen.

mesenteric artery (Fig. 3). At the aortic bifurcation, extension of the cystic lesion was identified in the right lower quadrant of the abdomen. These cystic structures were supplied by branches of the superior mesenteric artery (SMA) and celiac arteries. Oral contrast did not show communication between the cysts and the bowel. Portal venous drainage of the cysts was via the superior mesenteric vein. Malrotation of the bowel was seen, with the small bowel located on the right side and large bowel on the left, with the appendix in the left upper quadrant. No volvulus was evident. Some ascites was present. MRI confirmed the presence of a cystic left paravertebral structure consistent with myelocele and definite separation of the large thoracoabdominal enteric cyst from the myelocele (Fig. 4).

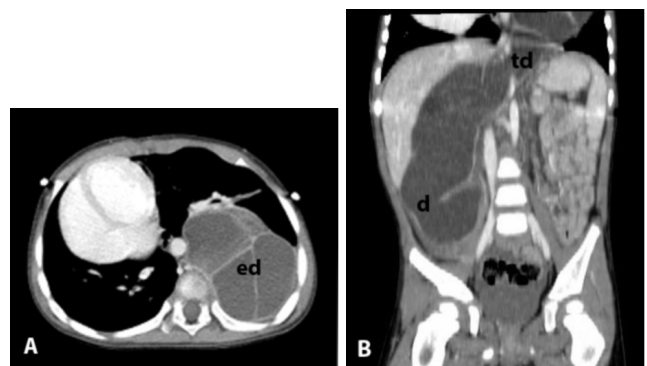


Figure 4. 2-year-old male with thoraco-abdominal enteric duplication cyst. A. Axial CT in lower chest demonstrates hypoplasia of left lung and dextroposition of heart related to enteric duplication (ed) with multiple folds in the chest. B. Coronal CT reformat demonstrates transdiaphragmatic duplication (td) showing entrance through diaphragm extending to right lower quadrant (d).

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Abdominal surgery revealed an abnormal bowel-like cystic mass within the jejunal mesentery that extended cephalad, crossing the aorta between the SMA and celiac axis. Intestinal malrotation with the entire bowel on the right side of the abdomen was also seen, with several duodenal kinks, and with the appendix and cecum were located in the left upper quadrant. Following separation of the cyst from the adjacent bowel wall, the malrotated bowel was placed appropriately in a nonrotated state, and the abdomen was closed. Thoracic surgery showed extension of the large thoracoabdominal cystic mass into the posterior mediastinum, consistent with the CT exam. This cyst extended to but was separate from what appeared to be a myelocoele at the apex of the left chest. The cystic mass was resected, and the myelocoele was then repaired, as was the diaphragmatic hernia defect.

Pathologic examination revealed that the specimen from the inferior and superior portion of the thoracoabdominal cyst consisted of two tubular structures resembling bowel (Fig. 5). The abdominal portion measured 10.0 cm in length and 3.0 cm in diameter at the widest portion. The thoracic portion of the cyst measured 24.5 x 3.5 cm, with the superior aspect of the thoracic cyst ending in a blind sac. Histology of the thoracoabdominal cyst resembled portions of alimentary tract, including small intestine, gastric antrum, and esophagus. The third specimen, excised from the spinal meninges, thought to be myelocoele, consisted of a rubbery cystic structure measuring 3.0 x 2.0 x 1.5 cm. Sections of this spinal cyst demonstrated a glial component and foregut elements consisting of bronchial cartilage and mucus glands, consistent with neurenteric cyst (Fig. 5).

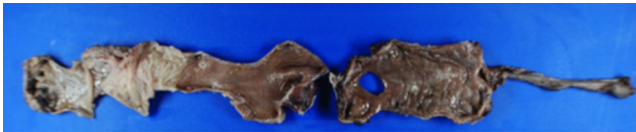


Figure 5A. 2-year-old male with thoraco-abdominal enteric duplication cyst. Duplication cyst, proximal on left side of figure. Pale proximal mucosa corresponds to esophageal lining. Gastric and small intestinal features are found sequentially in more distal portions.

Discussion

Gastrulation establishes the three germ layers of the embryo; it begins with formation of the primitive streak following notochord formation. Split-notochord syndrome results from failure of the three germ layers to form correctly, thus presenting various anomalies such as gastrointestinal duplication, neurenteric cyst, vertebral anomalies (hemivertebra, butterfly vertebra, and spina bifida), split-cord malformations (diplomelia, diastematomyelia), and subcutaneous malformations. The early failure of the notochord to completely separate from the foregut results in both the malformation of mediastinal foregut cyst and a vertebral cleft with an intraspinal neurenteric cyst. The

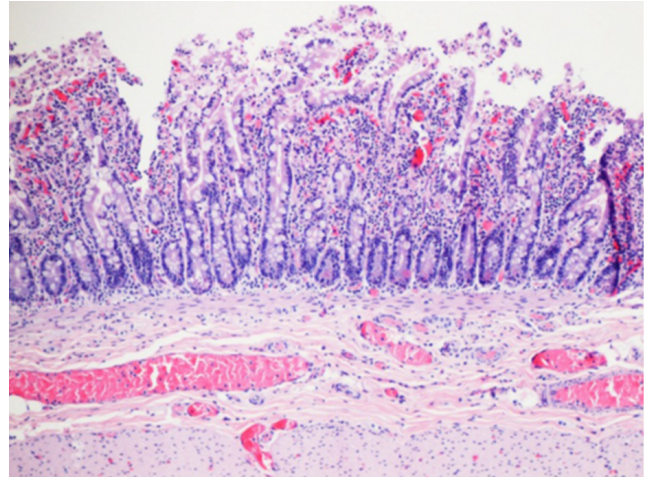


Figure 5B. 2-year-old male with thoraco-abdominal enteric duplication cyst. Duplication cyst. Small intestinal mucosa, submucosa, and muscularis propria. Original magnification, 100x; hematoxylin and eosin stain.

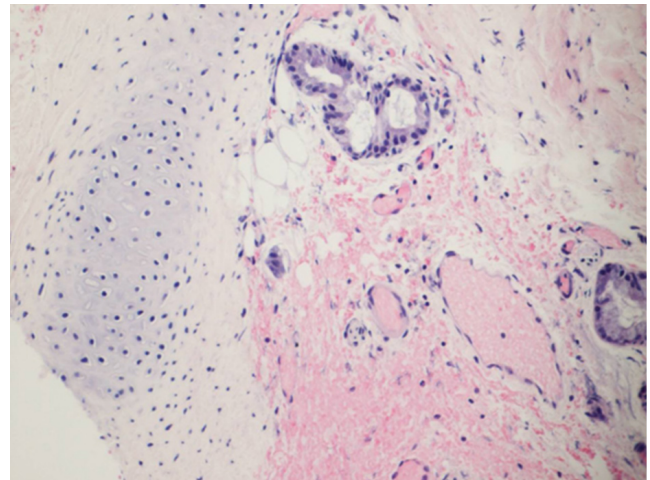


Figure 5C. 2-year-old male with thoraco-abdominal enteric duplication cyst. Neurenteric cyst. Foregut elements include hyaline bronchial cartilage and bronchial mucus glands. Glial elements (not shown) are also present. Original magnification 200x; hematoxylin and eosin stain.

duplication cysts usually move caudally as the embryo grows and the intrathoracic viscera descend. The vertebral anomalies often remain in the lower cervical spine (4, 5). Many of these patients have associated abdominal duplication cysts.

Gastrointestinal duplications can occur anywhere in the alimentary tract. The most common site is the ileum; next, in order, are the esophagus, jejunum, colon, stomach, and appendix. They usually occur on the mesenteric side and share blood supply with the intestine; occasionally they have a separate blood supply. The clinical presentation of alimentary tract duplications includes bleeding, abdominal pain, or intussusception, according to the nature of the

cysts and the site of origin. Intrathoracic duplication cysts can enlarge enough to cause cardiac and respiratory symptoms. Intestinal duplications are rarely found in association with malrotation, as was the case in our patient. Two cases in the literature included combination of duplications of bowel and malrotation (6). Thoracoabdominal duplications constitute approximately 2-4% of all duplications (7). They tend to be long, often large tubular lesions located in the posterior mediastinum, usually to the right of midline. In our case, the duplication was to the left of the midline in the chest. Closed at their cranial end, thoracoabdominal duplications pass through the diaphragmatic hiatus, where 60% of the lesions communicate with normal duodenum, jejunum, or ileum (8, 9). The remainder end blindly in the abdomen. No communication was seen in our patient of the blind ending cyst.

The neurenteric cyst is classically a solitary lesion in the cervical region, found less frequently in thoracic and lumbar locations. In some instances, the cysts may be attached or even communicate with the spinal canal; they are usually located anterior or anterolateral to the cord and are associated with vertebral and intraspinal abnormalities. Bone abnormalities, if present, are likely to involve the anterior column. These rare congenital disorders consist of a smooth muscle-walled structure containing mucosal epithelium, including ectopic gastric mucosa. Clinically, the syndrome is diagnosed whenever there is an association of mediastinal or abdominal cyst with malformation of the spinal cord and column.

In conclusion, cross-sectional imaging is important in planning surgery for the successful treatment of such a complex case.

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