Foregut Duplications

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Enteric duplication is a rare congenital anomaly with an incidence of approximately 1 in 4500 live births, with foregut duplications comprising approximately one-third of these. Duplications occur anywhere from the oropharynx to the anus; less than 3% are located in the neck, 60% are intrathoracic, 30% within the upper abdomen, and 5–10% thoraco-abdominal. Clinical presentation of these anomalies is highly variable with respect to size, location along the alimentary tract, and clinical progression. Many duplications have an unpredictable clinical course, which favors early resection. The spectrum of clinical significance from asymptomatic to sometimes growth restrictive and severe with the potential for associated anomalies emphasizes the importance of early diagnosis and excision. While most duplications are benign, ectopic tissue (gastric or pancreatic), associated anomalies, and malignant degeneration remain concerns.

Duplications can be cystic (80%) or tubular (20%) and are defined by three characteristics: (1) a well-developed smooth muscle within the cyst wall; (2) an epithelial lining of gastrointestinal origin, and (3) attachment to a portion of the alimentary tract (Table 49.1).

Although histologically the mucosa sometimes differs from the associated anatomic structure or location, duplication cysts are named for their location rather than their mucosal composition. Cystic duplications typically share a common wall and not a lumen, while tubular duplications often demonstrate luminal communication; both share a common blood supply with the structure from which they originate. Overall, duplications are most common in the abdomen (80%), with synchronous alimentary tract duplications present in up to 20% of patients. Approximately 85% of duplications are found within the first 2 years of life in some series, likely a product of enhanced imaging and diagnostic capabilities in the antenatal period, which can detect foregut duplications as early as 12 weeks. Many foregut cysts are found on routine imaging and are asymptomatic; however, the presentation of the symptomatic cyst depends on its anatomic level, the potential for creating a local mass effect, and function with regard to acid-secreting cells.

Table 49.1 Workup, differential diagnosis, and treatment of foregut duplications

Type	Diagnostic method	Differential diagnosis	10 treatment	20 or 30 treatment
Bronchogenic	CT/MRI	• Pulmonary sequestration • CPAM	Resection	None
Esophageal	CT	• Abscess • Leiomyoma	Resection	Marsupialization with mucosal stripping
Neurenteric	MRI		Resection	Laminectomy/laminotomy
Gastric	CT/US	• Abscess • Splenic cyst	Resection	Cyst gastrostomy with mucosal stripping
Duodenal	CT/MRCP	• Choledochal cyst • Pseudocyst	Resection	Cystduodenostomy with mucosal stripping, endoscopic unroofing, pancreaticoduodenectomy
Pancreatic	CT/MRCP	• Pseudocyst • Cystic neoplasm	Resection	Cystgastrostomy, cystjejunostomy, pancreaticoduodenectomy

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P. Mattei (ed.), *Fundamentals of Pediatric Surgery*, https://doi.org/10.1007/978-3-031-07524-7_49

Foregut duplications arise as a result of the anomalous formation of the foregut during development. The primitive foregut is the progenitor for the pharynx, respiratory tract, esophagus, stomach, and duodenum to the ampulla of Vater. The laryngotracheal groove first appears around the third week of gestation. The dorsal portion differentiates into the esophagus and the ventral portion into the respiratory tract. Given the common embryologic origin, the term foregut duplication encompasses all bronchogenic cysts, esophageal duplications, mixed lesions of esophageal and respiratory origin, and duplications of the stomach, duodenum, and pancreas. Several models have been proposed to explain the development of foregut duplications, however, there is no widely accepted theory. Aberrant luminal recanalization as a result of duplicate vacuolization of the solid cord, failure of regression of embryonic diverticula, abortive twinning, and environmental factors such as trauma or hypoxia have been hypothesized as causative factors. The split notochord theory for the formation of these cysts is supported by the presence of vertebral anomalies in some patients with thoracic and thoracoabdominal duplication cysts. In these cases, the notochord splits, allowing herniation of the endodermal gut, which may further interfere with the anterior fusion of the vertebral mesoderm. Vertebral anomalies such as scoliosis, hemivertebrae, and spina bifida are the most frequent associated malformations with foregut duplication; however, there is also an apparent association with malrotation, esophageal atresia, TEF, congenital diaphragmatic hernia, and cardiac/pericardial defects.

Thoracic Duplications

Bronchogenic and esophageal duplications comprise primary intrathoracic foregut cysts and are found most commonly within the posterior mediastinum. Esophageal duplications are more common and represent 20% of all foregut duplications. Thoracic duplications are more likely to be symptomatic at presentation due to airway or esophageal compression. They provoke a range of symptoms from relatively benign such as vomiting, cough, or shortness of breath to severe, including recurrent pneumonia and hemoptysis due to the presence of heterotopic tissue. Up to one-half of all esophageal duplication, cysts contain gastric or pancreatic mucosa, leading to bleeding, ulceration, perforation, or fistula as the presenting symptom. Esophageal duplications rarely demonstrate luminal communication or a shared muscular wall. Approximately 25% of esophageal duplications are cervical, typically present as an asymptomatic neck mass, and are commonly misdiagnosed as lymphangioma, thyroglossal duct or branchial cleft cysts, or solid tumors. Duplications of the mid esophagus represent 15–20% of esophageal duplications, and the remaining majority are found in the distal esophagus, where they are rarely symptomatic. In many cases, these cysts are also at risk for infectious complications and can present with fever and back or chest pain. The superimposed infection of these cysts can cause rapid expansion and subsequent symptomatic compression of the airway, esophagus, or mediastinal vasculature.

Chest radiograph during the initial workup for presenting symptoms will often reveal an intrathoracic foregut cyst as well as associated vertebral anomalies. While some cysts are radiographically well defined, given the high density of mucoid content, it is important to note that AP chest radiographs may not be adequate for identifying these cysts given the overlap of mediastinal structures. It is therefore recommended to obtain lateral views for more accurate identification. If vertebral anomalies are identified, and an associated duplication cyst is suspected, it is crucial to obtain cervical and thoracic MRI studies to identify potential neurenteric cysts, which have the potential to produce a range of neurologic symptoms from severe pain to paralysis due to extension into the spinal canal. If identified, prompt treatment is imperative to avoid the need for emergent laminectomy or laminotomy. Complete resection of neurenteric cysts can often be approached thoracoscopically but, depending upon the degree of spinal column extension, might require neurosurgical intervention or emergent decompression of the spinal cord. If emergent decompression is required, a delayed resection by thoracoscopy or thoracotomy is appropriate.

Bronchogenic and esophageal cysts are often identified by the antenatal US, with a characteristic echogenic internal ring and *double-wall* sign. If a symptomatic thoracic duplication cyst is suspected by either US or CXR, further workup with axial imaging is preferred, unless a neurenteric cyst is suspected. CT is useful for preoperative planning to define relationships to surrounding structures, characterize the nature of the cyst, and identify intraluminal communication. Intravenous contrast may help to identify proximity to vascular structures, but duplications will not enhance. The cyst wall is often calcified and rim enhancing and therefore commonly confused for an abscess; however, in the absence of clinical symptoms, this diagnosis is unlikely. Some describe using a technetium scan to document ectopic gastric mucosa; however, this is not essential for operative planning or treatment. Both chest and abdominal CT should be included in the initial workup, as approximately 25% of patients with a thoracic duplication have a synchronous abdominal enteric duplication.

Our preferred method of resection for both esophageal and bronchogenic cysts is thoracoscopy on the side of the lesion or from the right for posterior mediastinal duplications. A left-sided bronchogenic cyst in the pulmonary hilum might demonstrate a long segment of attachment to the left pulmonary artery (Fig. 49.1). Preoperative antibiot-

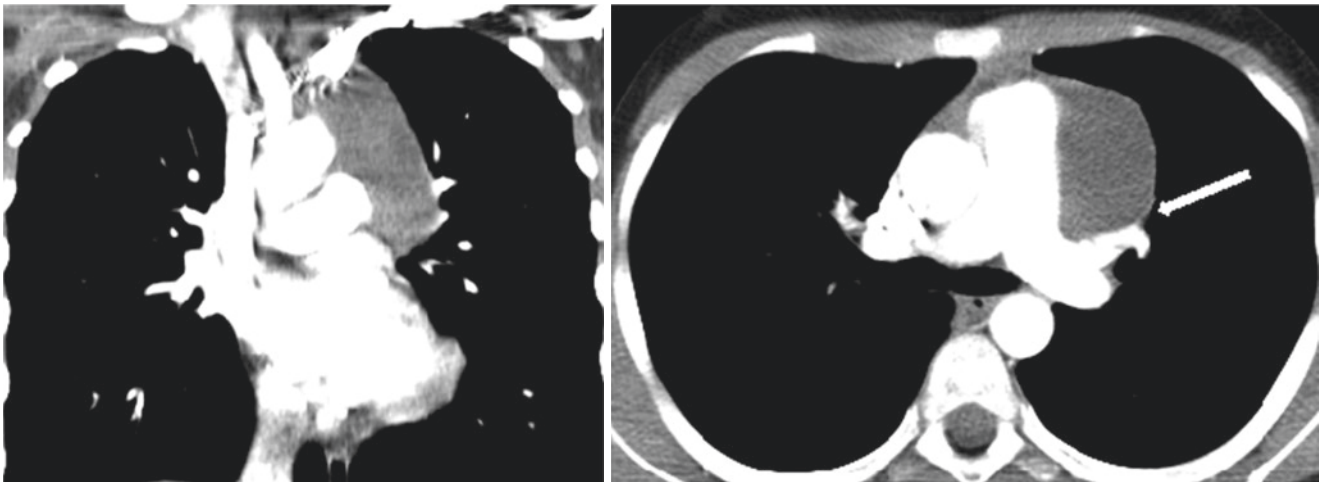

Fig. 49.1 CT of the chest showing bronchogenic cyst near the left pulmonary hilum. The arrow demonstrates the long segment where the lesion was adherent to the left pulmonary artery requiring slow thoracoscopic dissection

ics are given to all patients. An attempt is always made for complete resection without violation of the esophageal or bronchial lumen. If there is an extensive bed of dissection from which drainage is expected, a soft 10 or 15Fr round closed-suction drain is left in place and can typically be removed on the first postoperative day. Standard chest tubes are never required. If the duplication is intimately associated with the esophagus, careful inspection with the passage of a bougie is usually adequate to identify a full-thickness defect—neither routine esophagram nor chest drain is usually necessary. If the esophagus is entered and requires closure, a drain should be considered, and an esophagram before initiating a diet is prudent, though most surgical esophageal leaks can safely be managed conservatively. If resection cannot be safely completed, as much of the cyst as is safely accessible is resected and the mucosa is stripped from the unresectable portion of the lesion. In this situation, dissection can be performed with electrocautery through the submucosal plane. If this is too difficult or incomplete, the remaining mucosa can be destroyed with electrocautery or Argon-beam coagulation.

Occasionally a cyst in the central mediastinum is confined by the great vessels, shielding it from either a right- or left-sided thoracoscopic approach. These lesions sometimes require median sternotomy in conjunction with a cardiac surgeon (Fig. 49.2). After the lesion is freed up, we retract the vessels to gain exposure; however, if the blood pressure suddenly drops, requiring the release of the retractor, the dissection needs to be completed in small intervals. A small defect in the anterior trachea is sometimes evident after resection where a small common wall existed, but can usually be closed with an absorbable monofilament suture.

Multiple large series have proven the safety of thoracoscopic resection of these cysts, with the most common complications similar to those found in other thoracic interventions. Patients who are symptomatic preoperatively are more likely to have a complication and a longer length of stay due mostly to pain control. Of note, an infection may cause rapid enlargement of a duplication, likely provoking symptoms in an otherwise newly diagnosed or known duplication that was previously asymptomatic. This is of heightened importance for duplications close to the airway for both intubation and operative planning. Intraoperatively, specific attention to remaining close to the cyst wall during resection is pertinent. Vascular supply is almost uniformly matched by the associated anatomy, with an extremely rare probability of ectopic vascularization. Given that most of

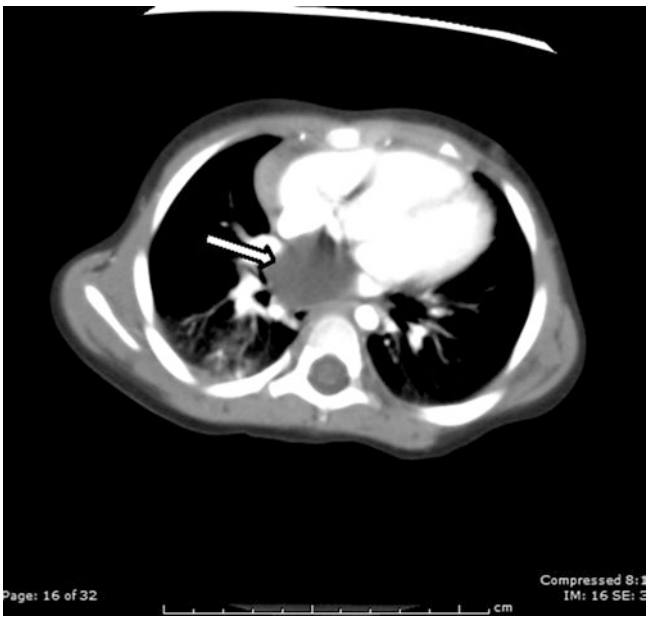

Fig. 49.2 CT demonstrating a central bronchogenic cyst where no safe window for resection exists from either side thus a median sternotomy was performed

these cysts do not demonstrate luminal communication, remaining close to the cyst wall during resection decreases the likelihood of inadvertent esophageal or tracheal perforation.

Thoracoabdominal cysts comprise approximately 2% of all duplications and communicate with the distal alimentary tract through the esophageal hiatus in 60% of cases. These lesions can present with severe respiratory distress in the neonate due to fluid accumulation in the cyst and compression of thoracic structures due to lack of luminal communication with thoracic foregut structures. CT is important for preoperative planning as with all foregut cysts. Our preferred approach is combined laparoscopy and thoracoscopy with the closure of the diaphragmatic defect. We favor a one-stage resection, while in multiple series, interval abdominal and thoracic intervention is described; there is currently no data to support a superior approach. Thoracoabdominal cyst resections are more likely to be incompletely resected due to this combined intervention, with few reports of a missed thoracic cyst component.

Abdominal Duplications

Abdominal foregut duplications represent approximately 15% of all duplications. Of these, gastric duplications comprise 5% and have a unique presentation as they are palpable on physical examination as a mobile epigastric mass in up to 50% of patients. Gastric duplications rarely communicate with the gastric lumen and in most cases are found on the greater curvature with occasional retroperitoneal extension or isolated retroperitoneal development. Due to the retroperitoneal or posterior location of some gastric duplications, the most common misdiagnosis is pancreatic pseudocyst. They provide the highest risk for intestinal and gastric obstruction given their size and present with symptoms of poor feeding, vomiting, and reflux, often initially diagnosed as GERD or hypertrophic pyloric stenosis due. There is a strong predilection for active heterotopic tissue and it is not uncommon for the initial presentation to be perforation with hematemesis and melena. Preferred workup for these masses remains CT, but in many cases, the diagnosis may be suspected on a UGI completed for symptoms consistent with reflux. One advantage of CT is the utility of oral contrast to identify communication with the gastric lumen. The goal of surgery is complete resection and, in some cases, may require reconstruction if the lumen is violated. Laparoscopic excision is our preferred approach. Most gastric duplications share a common blood supply with the stomach through the gastroepiploic vessels. Duplications with heterotopic gastric tissue provide for a more difficult dissection due to recurrent inflammation from acid production. If a safe plane cannot be established between the duplication and the native gastric wall, a partial gastrectomy can be performed by stapling off a wedge of the stomach where the duplication is attached. The lesion seen in Fig. 49.3 was resected by identifying the plane separating the gastric wall from the cyst (Fig. 49.4). Untreated gastric duplications are most often associated with malignant degen-

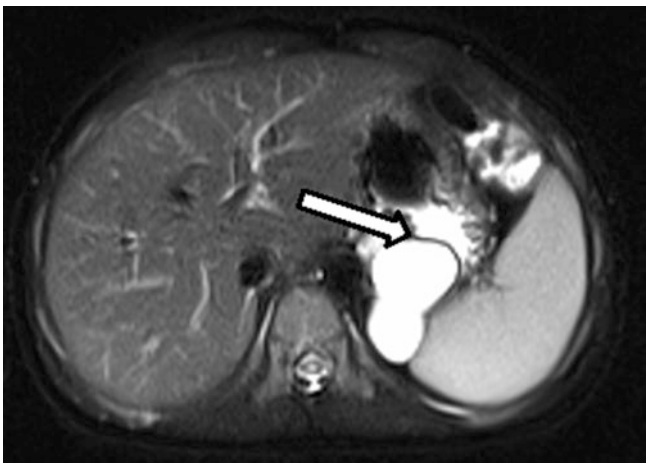

Fig. 49.3 MR showing enteric duplication intimately associated with the stomach as demonstrated by the arrow

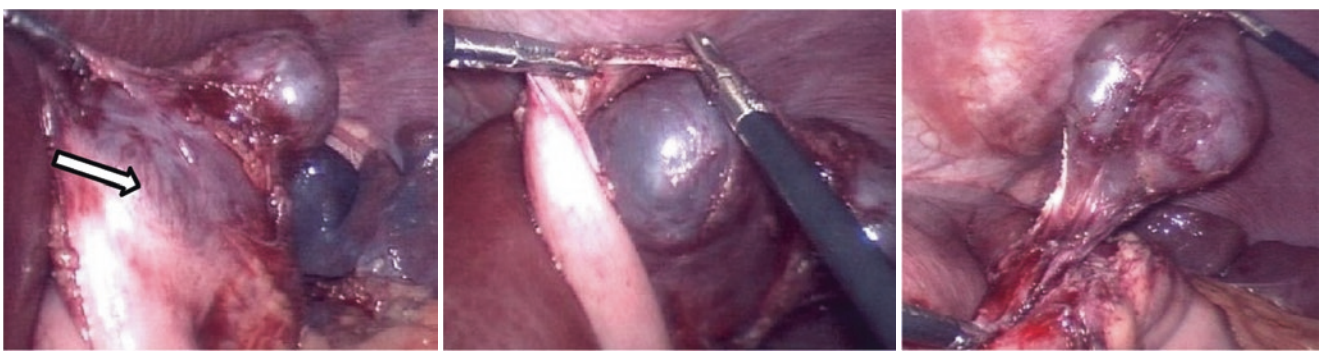

Fig. 49.4 Laparoscopic excision of a gastric duplication demonstrating the initial appearance with a subtle transition from stomach to cyst (arrow) then the subsequent dissection, which identifies the plane to allow simple complete resection

eration due to the high prevalence of gastric tissue and lack of luminal communication resulting in chronic inflammation and metaplasia.

Duodenal duplications represent up to 5% of intestinal duplications and are most commonly found in the posteromedial aspect of the second and third portions of the duodenum, with luminal communication in approximately 25% of cases. A majority of these cysts are found on the mesenteric side of the anterior bowel wall. Given the proximity to the pancreaticobiliary system, patients with duodenal duplications are at higher risk for hyperbilirubinemia and jaundice, and pancreatitis from the mass effect of these lesions prompting frequent misdiagnosis as a choledochal cyst. Additionally, heterotopic gastric tissue is common with these cysts and presents a greater risk for peptic ulceration and subsequent associated complications. The most common presentation is early satiety, vague abdominal pain, vomiting, and failure to thrive. If a duodenal duplication is suspected, workup with CT or MRCP is appropriate to determine the relationship to the pancreatic and biliary ducts. Our preferred surgical approach is laparoscopic resection, with intraoperative cholangiography available to ensure the integrity of the biliary and pancreatic ductal systems both during and after resection of the duplication. In the setting of duodenal resection, UGI is performed prior to the initiation of PO intake. Complex or highly morbid resections are avoided by performing internal drainage and mucosal stripping.

Pancreatic duplications are rare and follow a similar vague presentation pattern of most foregut duplications. Pancreatic head duplication cysts are the most common of this subgroup (50%), with the remainder in equal distribution between the body and tail. Workup for these lesions again includes CT or MRCP as the definition of the ductal systems and proximity of the cyst is pertinent to appropriate management and the goal of complete resection. Diagnosis of these cysts is difficult due to similarities with pancreatic pseudocysts on imaging but are favored in the differential due to the timing of diagnosis, typically antenatally up to 6 months of life. Body and tail cysts can be appropriately managed with distal pancreatectomy with splenic preservation. However, pancreatic head duplication cysts provide a unique challenge and dictate an alternative surgical approach that emphasizes enucleation over complete resection due to the likely intimate proximity and possible communication with pancreatic and biliary ductal structures. Knowledge of the ductal anatomy is crucial to the appropriate management of these lesions. Internal drainage and pancreaticoduodenectomy are secondary options if enucleation is not feasible or safe due to the involvement of biliary or pancreatic ductal structures. Frozen sections will demonstrate smooth muscle lining, distinguishing them from pancreatic pseudocysts. After pancreas resection, drain placement is recommended.

Antenatal Diagnosis

A separate discussion of prenatal diagnosis of foregut duplications is pertinent given their unique pathology that may be appropriately diagnosed with routine antenatal screening, with a prenatal diagnosis rate of close to 50%. Detection as early as 12 weeks is possible and typically observed by 20 weeks. There are two distinct findings on US that suggest a duplication: a double-wall sign (an inner hyperechoic layer of mucosa-submucosa and an outer hypoechoic layer consisting of muscularis propria) and peristalsis of the cyst wall. Once identified, serial US should be performed as the growth of these cysts can cause complications requiring fetal intervention. Hydrops related to foregut cysts, primarily bronchogenic and esophageal, have, in rare cases, required thoraco-amniotic shunting. Additional characterization of foregut duplications can be achieved with fetal MRI and is recommended in conjunction with echocardiography. Associated anomalies have been reported in up to 50%. Close follow-up and elective resection are recommended by 6 months of age given the association with malrotation and risk of volvulus or obstruction. It is important to emphasize the variable nature of these duplications in terms of size, symptoms, and location. Approximately 80% of foregut duplications will become symptomatic and there are currently no predictive factors to determine the timing for symptomatic presentation or which patients will experience the most severe complications. Therefore, early elective resection should be strongly considered in all patients with identified foregut duplications.

Concerns and Complications

Pertinent concerns and complications with foregut duplication cysts remain the risk of severe symptoms, complications from their expansion, and the potential for malignant degeneration if left untreated or incompletely excised. Expectant management is never recommended. Diagnosis relies on prompt imaging, most commonly CT or MRI, whenever a duplication is suspected. MRI avoids ionizing radiation and is the best modality for imaging concomitant spinal abnormalities. The drawback of MRI is that young children often require general anesthesia or sedation. CT can provide detailed imaging of the bronchial tree and its relationship to the lesion and, when performed with intravenous contrast, characterize the vascularity of the lesion. An upper GI contrast study provides information about the connection of duplication to the enteric system, but this information is not vital prior to surgical intervention. All patients with a foregut duplication should have a diligent search for associated anomalies and other duplications. US is of little utility in thoracic foregut duplications except in neonates and although

usually diagnostic, cysts adherent to the duodenum or pancreas will likely require further imaging in the form of MRCP, CT, or ERCP. Technetium-99 m pertechnetate scans might be useful in the setting of small duplications with heterotopic gastric mucosa, but if duplications can be localized by other means, then resection is the treatment with or without ectopic gastric mucosa. Endoscopic US is effective for the diagnosis of foregut cysts in adults but there is little experience in children.

The primary concern remains malignant degeneration and, although rare, is associated with a very poor prognosis. Given that approximately 80% of duplications are symptomatic at the time of surgery, it is unlikely for a patient to have a quiescent duplication cyst, whether symptomatic from mass effect or intrinsic activity. Partial resection is not recommended in any case due to high recurrence rates. If complete resection is not feasible due to an anatomic location prohibiting safe resection without significant morbidity, mucosectomy is an acceptable option especially for removing acid-secreting tissue. However, surveillance of these patients should be more frequent with additional resection, if necessary. With mucosectomy, there remains a high risk for recurrence and potential remnant tissue that can undergo malignant degeneration because of the retained pancreatic or gastric mucosa. For lesions not amenable to resection or mucosectomy due to anatomic location, drainage procedures such as cyst-gastrostomy, cyst-duodenostomy, or Roux-en-Y-cyst-jejunostomy may be considered, with a plan for lifelong surveillance, repeat imaging, and biopsy. Minimally invasive complete resection of foregut cysts remains our recommended and favored approach and is associated with exceptional outcomes, low complication rate, and very low recurrence.

Editor's Comments

Foregut duplications in the chest include bronchogenic cysts and esophageal duplications. Nearly all can and should be excised thoracoscopically. Exceptions include some that communicate with or involve a long segment of the esophagus or airway and those that were infected at one time and are therefore likely to be extremely adherent. As with all operations performed in the mediastinum, careful attention to avoid injury to adjacent structures (phrenic nerve, vagus, thoracic duct) is paramount, though fortunately these lesions typically separate from important structures with gentle blunt dissection, until you reach the organ of origin, when patience and meticulous technique are critical.

Spillage of the sterile contents of the cyst does not appear to increase the risk of recurrence or infection and is often performed deliberately to aid in the dissection of a large cyst or to remove it from the chest. When dealing with an especially adherent or fibrotic cyst wall, it is acceptable to enter the lumen and completely strip the mucosa, leaving the fibrotic wall behind, but it is especially important to identify and repair communication with the lumen. Except when a lumen has been breached or frank infection is present, routine placement of a drain or chest tube is unnecessary.

Long, tubular thoracoabdominal esophageal duplications can also be approached thoracoscopically, by mobilizing the thoracic portion first and then completing the dissection in the abdomen by laparoscopy or laparotomy. Extensive preoperative imaging with three-dimensional reconstruction is very useful in planning these challenging operations. Because retained mucosa places the patient at lifelong risk of complications and cancer, every effort should be made to excise it, though it is probably safe to leave the outer fibrous wall. In fact, with esophageal duplications, one can sometimes split the shared esophageal muscle coat and simply enucleate the mucosal cyst itself. Marsupialization or creation of an enteric communication is not a definitive treatment and should only be done under extreme circumstances, most commonly with duodenal duplications in which the only reasonable alternative is to perform a Whipple. It is probably impossible to completely destroy an enteric epithelium with an energy source, including the argon-beam coagulator; this should not be considered a definitive treatment of an enteric duplication.

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