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Surgical management of critical congenital malformations in the delivery room

Anthony Ferrantella, Henri R. Ford, Juan E. Sola*

Department of Surgery, Division of Pediatric Surgery, University of Miami, Miami, FL, USA

A B S T R A C T

Newborn emergencies that occur in the delivery room are frequently the result of life-threatening congenital anomalies that can result in death or severe disability if not treated in the immediate postnatal period. Prompt recognition and treatment of such disorders are paramount to ensuring the wellbeing of the infant. As congenital anomalies are frequently being diagnosed earlier due to improved prenatal detection, the coordination of planned interventions for life-threatening malformations is also becoming more common. This article serves as a guide for the presentation and initial management of the most common non-cardiac, newborn surgical emergencies.

1. Introduction

Various life-threatening congenital anomalies may present in the delivery room or shortly thereafter. These anomalies require prompt recognition by both obstetricians and pediatricians given that delayed intervention may result in permanent disability or even death. With recent advances in prenatal detection of congenital malformations over the past several decades, many of these disorders are routinely detected by fetal ultrasound, thereby alerting clinicians early to their presence. In such cases, consultation with an experienced team including perinatologist and pediatric surgeon should take place as early as possible to ensure the availability of specialized personnel and resources to appropriately intervene on these potentially life-threatening malformations in the delivery room. The following summary provides an overview of the most common non-cardiac congenital anomalies that require immediate intervention following birth.

2. Airway obstruction

2.1. Pierre Robin sequence

Pierre Robin sequence may occur in isolation or as part of several other congenital anomaly complexes and syndromes. Although this condition has multiple etiologies, its hallmark is the presence of a recessed, hypoplastic mandible. This leads to posterior displacement of the tongue causing upper airway obstruction. Additionally, the displacement can prevent fusion of the palatal shelves during early gestation, which results in a cleft palate in the majority of cases [1]. Airway compromise can lead to hypoxia, cardiopulmonary arrest, pulmonary hypertension, and failure to thrive. Affected patients should be monitored to detect apnea and airway obstruction. Prone positioning is helpful to minimize airway obstruction, and insertion of a nasopharyngeal trumpet can protect the airway. In some rare cases, airway obstruction may be refractory to these measures and require endotracheal intubation or tracheostomy. Initially, feeding difficulties are also common and may necessitate placement of a nasogastric feeding tube and eventual surgical gastrostomy tube [1,2]. A trial of non-operative measures should be utilized in these patients as with time most infants are able to keep their tongue away from the larynx and mandibular growth during the first year of life allows for resolution of airway problems in a majority of patients. Other surgical options include distraction of the mandible, in which fracture of the mandible is performed and an external fixator is used to open the oral passage. This approach has the benefit of permanently changing mandibular size to create adequate space for the tongue, allowing many patients to avoid tracheostomy altogether when performed early [3].

2.2. Congenital high airway obstruction syndrome (CHAOS)

Congenital high airway obstruction syndrome (CHAOS) is characterized by complete or near-complete upper airway obstruction due to atresia, membranous webs, or stenosis of the larynx or trachea (Fig. 1). Cases of atresia present with asphyxia at the time of birth and require emergent tracheostomy. The diagnosis can be made prenatally via ultrasonographic findings of large echogenic lungs, dilated distal trachea, and flattened hemidiaphragms. If diagnosed prenatally, an Ex

* Corresponding author. Division of Pediatric Surgery, DeWitt Daughtry Family Department of Surgery, University of Miami Leonard M. Miller School of Medicine, 1120 NW 14th Street, Miami, FL, 33136, USA.
E-mail address: jsola@med.miami.edu (J.E. Sola).

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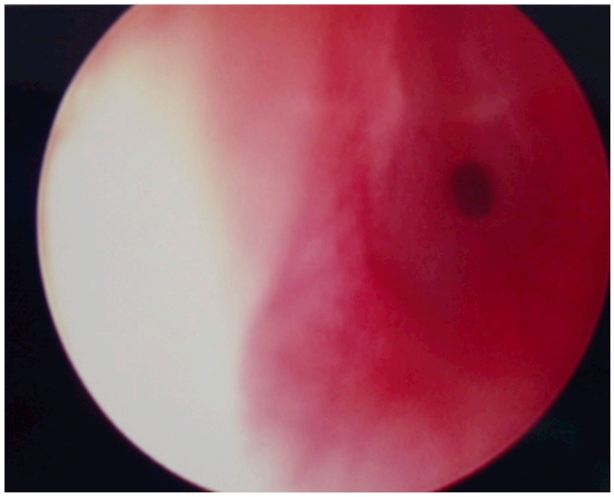

Fig. 1. Bronchoscopic view of trachea in infant with CHAOS diagnosed at birth and treated with emergent bedside tracheostomy. Patient also had associated type C esophageal atresia repaired during first week of life and eventually underwent tracheal reconstruction at a year of age.

Utero Intrapartum Treatment (EXIT) procedure with tracheostomy must be planned. Patients with webs or stenosis usually present during infancy with respiratory distress, stridor, and a weak or high-pitched cry. Depending on the degree of airway obstruction, some incomplete webs may be treated with simple lysis using endoscopic laser or sharp dissection, whereas more severe webs often require laryngeal reconstruction and tracheostomy [4]. Similarly, minor subglottic stenosis may be treated with an anterior cricoid-split or endoscopic division of cartilage with balloon dilation [5]. Congenital stenosis typically improves as the larynx grows, and fewer than half of children require tracheostomy.

2.3. Lymphatic malformations (cystic hygroma and lymphangioma)

Lymphatic malformations are benign lesions composed of dilated lymphatic channels or cysts. They are classified as macrocystic (previously cystic hygroma), microcystic (formerly lymphangioma), mixed or lymphovascular [6]. Macrocystic lymphatic malformations are composed of collections of large, interconnected lymphatic cysts lined by a thin endothelium. They present at birth as large, poorly delineated, translucent, soft masses covered by normal skin and are most commonly located in the cervicofacial region, axilla, or lateral chest wall (Fig. 2). They can be detected on prenatal ultrasound due to increased nuchal translucency and approximately half of cases are associated with chromosomal abnormalities such as Down syndrome. In contrast, microcystic lymphatic malformations may be present at birth or appear in the first few years of life. They are most frequently located in the proximal extremities, trunk, axilla, and oral cavity and present as a cluster of clear, translucent or hemorrhagic vesicles that can intermittently leak lymphatic fluid.

Although sclerotherapy is often successful in reducing the size of macrocytic lesions [7], surgical intervention may be needed at the time of birth if there is concern for airway compromise. Large neck lesions that are diagnosed prenatally may require an EXIT procedure in order to immediately secure the airway. If endotracheal intubation or tracheostomy is not possible, cannulation for extracorporeal membrane oxygenation (ECMO) may be required (Fig. 3). For lesions near the airway, an experienced pediatric surgeon should be consulted prenatally and be immediately available at the time of delivery.

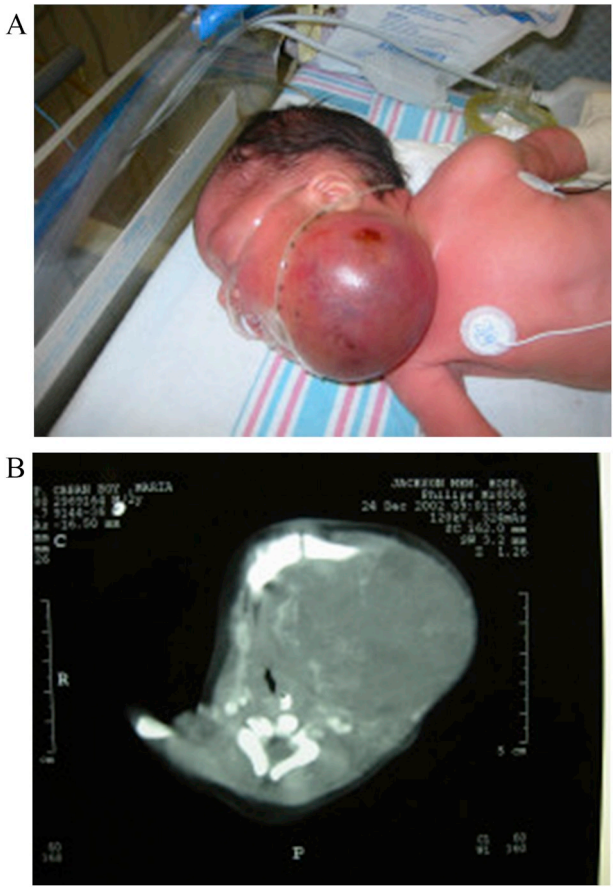

Fig. 2. Large lymphatic malformation in newborn infant (A). Computed tomography scan demonstrating compression of trachea (B).

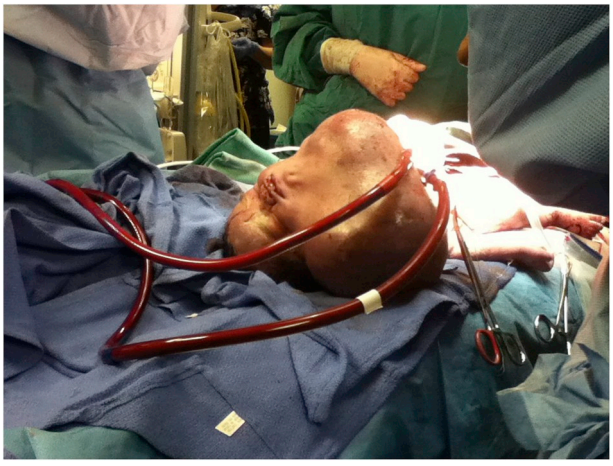

Fig. 3. Newborn infant with giant lympho-venous malformation treated with EXIT procedure for ECMO cannulation prior to surgical resection.

2.4. Mediastinal masses

Compression of the intrathoracic trachea or bronchi can also occur due to the presence of a mediastinal mass. A high-grade partial obstruction will result in air-trapping during exhalation with an over distended pulmonary lobe, whereas complete obstruction produces atelectasis. Compressive symptoms typically consist of tachypnea, wheezing, stridor, and cough. Teratomas of the anterior mediastinum are the most common cause in newborns [8]. These typically benign lesions can cause life-threatening respiratory distress requiring urgent

surgical excision. The presence of calcifications on imaging studies is diagnostic for mediastinal teratoma. Additionally, lymphatic malformations can occasionally be located in the mediastinum (5%) and cause mass effect with compressive symptoms.

2.5. Double aortic arch

In double aortic arch, the paired aortic arch arteries fail to remodel appropriately, which results in two aortic arches completely encircling the trachea and esophagus prior to rejoining posteriorly to form the descending aorta. The most common presenting symptoms are due to tracheal compression and include stridor, wheezing, cough, and respiratory distress [9]. Esophageal symptoms are also common and include difficulty feeding and vomiting. Clinicians must have a high index of suspicion given the rarity of double aortic arch, and the diagnosis can usually be confirmed by echocardiography. Treatment involves thoracotomy, or video-assisted thoracoscopy (VATS) where available, with ligation and division of the smaller aortic arch and patent ductus arteriosus.

3. Bronchopulmonary malformations

3.1. Bronchogenic cysts

Bronchogenic cysts arise from anomalous budding during development and can occur throughout the tracheobronchial tree but are most commonly located near the carina [10]. Although rare, these lesions are among the most common lower respiratory tract malformations. Affected patients typically present as older children with recurrent pneumonia, but they may become symptomatic during infancy. Newborns with rapidly enlarging central cysts can develop compressive symptoms consisting of respiratory distress, cyanosis, and feeding difficulty that necessitate early surgical excision (Fig. 4).

3.2. Congenital pulmonary airway malformation

Congenital pulmonary airway malformation (CPAM), previously known as congenital cystic adenomatoid malformation (CCAM), is the most common developmental anomaly of the lung [11]. Affected patients may present with respiratory distress in the newborn period. These lesions are often detected during routine prenatal ultrasound. Twenty-five percent of newborns with a prenatal diagnosis of CPAM are symptomatic at birth, and the likelihood of respiratory distress increases with the size of the lesion [12]. Fetuses with large CPAMs and/or hydrops have an especially poor prognosis. In these cases, treatment options include antenatal corticosteroids, drainage procedures, fetal surgery, or early delivery. In the postnatal period, the diagnosis of CPAM in a newborn with respiratory distress may be suggested by a chest radiograph demonstrating a hyperlucent lobe with midline shift. However, plain radiographs can often fail to detect these lesions. Therefore, all symptomatic newborns with a prenatal diagnosis of CPAM should also undergo advanced thoracic imaging with contrast-enhanced computed tomography (CT) or magnetic resonance imaging (MRI). For asymptomatic patients, delayed imaging can be obtained prior to elective resection of the lesion. Surgical resection is curative and has few complications. Lobectomy is generally preferred to wedge resection to ensure complete excision since the extent of some lesions may be difficult to identify intraoperatively.

3.3. Congenital lobar emphysema

Congenital lobar emphysema (CLE) results from progressive lobar hyperinflation due to a variety of disruptions in bronchopulmonary development. Although a definitive cause cannot be identified in approximately half of cases, the most frequently identified cause is obstruction of the developing airway, which occurs in 25% of cases [13].

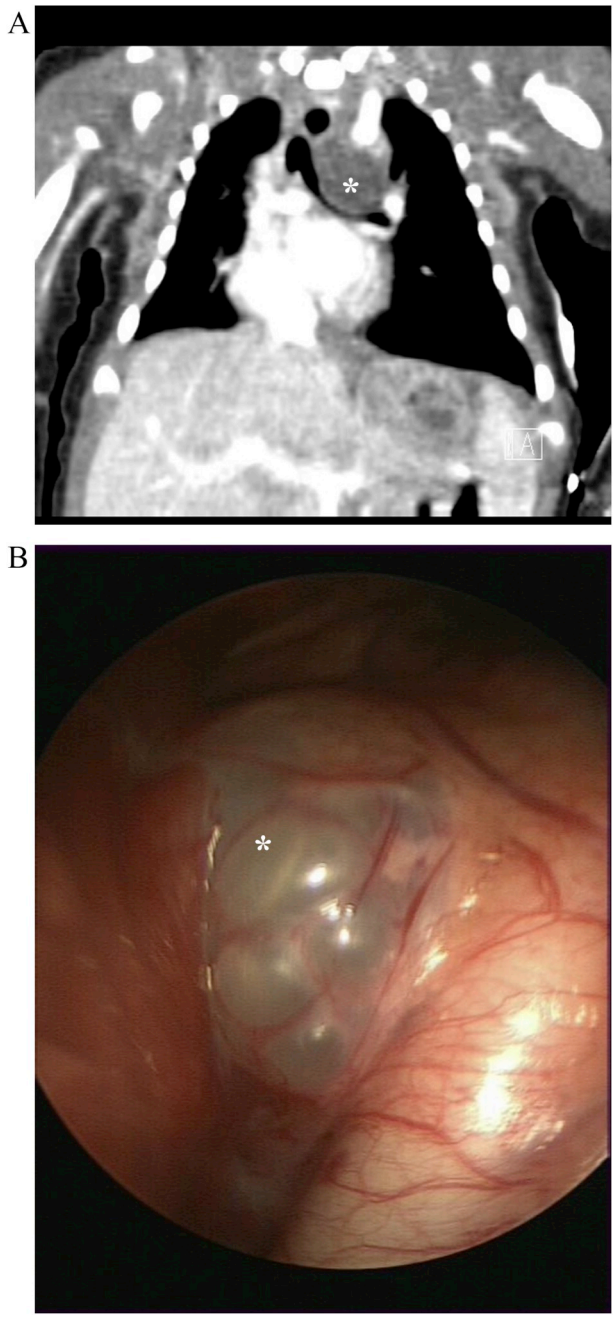

Fig. 4. Computed tomography scan of patient with bronchogenic cyst causing compression (*) of left mainstem bronchus (A). Thoracoscopic view of bronchogenic cyst (*) medial to aortic arch during resection (B).

This airway obstruction can be intrinsic or extrinsic, leading to the creation of a ball-valve mechanism of air trapping and hyperinflation. CLE is characterized by overdistention of one or more lobes of the lung. This leads to compression of the remaining lung tissue and potentially mediastinal shift. Affected infants are usually symptomatic in the neonatal period, with approximately 25–33% of cases presenting at birth, 50% by one month of age, and nearly all by six months of age [14]. Progressive respiratory distress develops rapidly in some infants, while others have a more gradual onset. The severity depends on the size of the affected lobe, the degree of compression of the surrounding lung tissue, and the extent of mediastinal shift [15]. The diagnosis can often be made from the characteristic appearance on chest radiograph, which typically demonstrates distension of the affected lobe, flattening of the ipsilateral hemidiaphragm, mediastinal shift, and compression of the

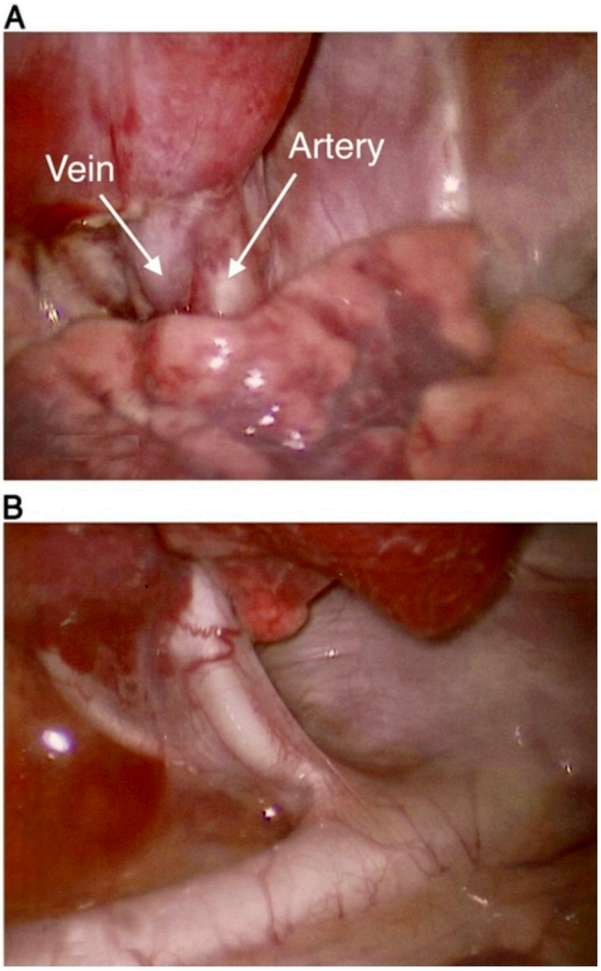

Fig. 5. Thoracoscopic view of left extralobar (A) and right intralobar (B) pulmonary sequestrations (superior to vessels). Used with permissions.

contralateral lung. CT or MRI may help establish the diagnosis and may demonstrate a source of airway obstruction. If the chest radiograph is obtained immediately after birth, the affected lobe may appear opacified due to retained fetal lung fluid. As the fluid is absorbed and the lobe becomes filled with air, progressive hyperinflation develops. CLE is sometimes detected by prenatal ultrasound but is often misdiagnosed as CPAM or bronchopulmonary sequestration [16]. Predictors of severe respiratory distress include polyhydramnios, fetal hydrops, and a decreased lung to thorax transverse area ratio (L/T value) [17]. The appropriate treatment of CLE in newborns with respiratory distress is surgical resection of the affected lobe.

3.4. Bronchopulmonary sequestration

Bronchopulmonary sequestration (BPS), also referred to simply as pulmonary sequestration, is a congenital abnormality of the lower airway that consists of a nonfunctioning mass of lung tissue. Although these lesions contain airway and alveolar elements, they lack normal communication with the tracheobronchial tree and receive their arterial blood supply from the systemic circulation (Fig. 5), which is of importance for distinguishing them from other lesions and planning their surgical excision [18]. The subtypes of BPS include intralobar sequestration, extralobar sequestration, hybrid BPS/CPAM lesions, and bronchopulmonary-foregut malformation. The most common subtype, intralobar sequestration, is located within a normal lobe and lacks its own visceral pleura. It typically has no bronchial connection to the proximal airway, but interalveolar connections (pores of Kohn) with the adjacent normal lung tissue can allow for the translocation of bacteria into the sequestration and cause recurrent infections, which is common for intralobar sequestrations. On the other hand, extralobar sequestrations are located outside of the normal lung and have their own visceral pleura, thereby lacking any intrapulmonary connections and rarely associated with infectious complications. Extralobar lesions can occasionally be extrathoracic located below the diaphragm in the retroperitoneum. Given that CPAM is the most common congenital anomaly of the lower airway, hybrid BPS/CPAM lesions comprise a substantial proportion of BPS [19]. These lesions have a blood supply from a systemic artery consistent with BPS, but they also have histologic features of CPAM. Bronchopulmonary-foregut malformation is a term used to refer to rare variants of BPS in which the sequestered lung tissue is connected to the gastrointestinal tract [20]. Many cases of BPS are initially detected by routine prenatal ultrasound, but may be difficult or impossible to distinguish from CPAM unless a systemic arterial blood supply can be identified. Ultimately, the final definitive diagnosis is only made by pathologic examination after surgical resection.

Most affected newborns are asymptomatic, but when symptomatic, BPS usually presents with respiratory distress at birth or shortly thereafter. Symptoms are more likely if the lesion is large, limiting the proper expansion of the adjacent normal lung. Later presentations are typically due to recurrent infections. For symptomatic newborns, those with a large lesion (occupying $\geq$20% of hemithorax) identified on ultrasound or plain radiographs, or those with risk factors for pleuropulmonary blastoma, early advanced cross-sectional imaging with CT or MRI is needed to confirm the diagnosis and define the aberrant vascular supply for preoperative planning. Early surgical resection of the affected lobe is indicated for these infants. For asymptomatic newborns without high-risk features, advanced thoracic imaging can be delayed up to six month of age to confirm the diagnosis and evaluate the need for elective surgical intervention [21]. Based on our institutional experience, we have developed and published a bronchopulmonary malformation surgical algorithm (Fig. 6) for the management of these malformations [22].

4. Esophageal atresia and tracheoesophageal fistula

Esophageal atresia (EA) and tracheoesophageal fistula (TEF) comprise a spectrum of relatively common congenital anomalies that typically occur together and are classified by their anatomic configuration. By far the most common form is type C, occurring in over 80% of cases. It consists of a blind proximal esophageal pouch and a distal TEF connecting the distal esophagus to the trachea [23]. The second most common variant is a pure EA, or type A, without any associated TEF. The other variants, in decreasing order of frequency, include TEF without EA (type E or “H-type”), EA with both a proximal TEF and a distal TEF (type D), and EA with only a proximal TEF (type B). The clinical presentation of TEF depends upon the presence or absence of EA. Given that EA is present in the vast majority of cases, polyhydramnios occurs in most pregnancies [24]. Nevertheless, many cases are not detected prenatally.

The majority of newborns with EA become symptomatic immediately after birth due to pooling of oral secretions resulting in excessive drooling, choking, respiratory distress, and inability to feed. A fistula between the trachea and distal esophagus leads to gastric distention and can result in aspiration pneumonia due to reflux of gastric contents through the TEF. The diagnosis of EA can be made by the inability to pass a nasogastric tube into the stomach and coiling of the catheter in the upper esophageal pouch, demonstrated on plain radiograph. A distal TEF may be suspected if radiographs also reveal a gas-filled gastrointestinal tract, whereas complete absence of intra-abdominal gas suggests the presence of EA without distal TEF. When the diagnosis is uncertain or a proximal TEF is suspected, a small amount of water-soluble, non-ionic, iso-osmolar contrast medium placed into the proximal esophageal pouch under fluoroscopic guidance will confirm

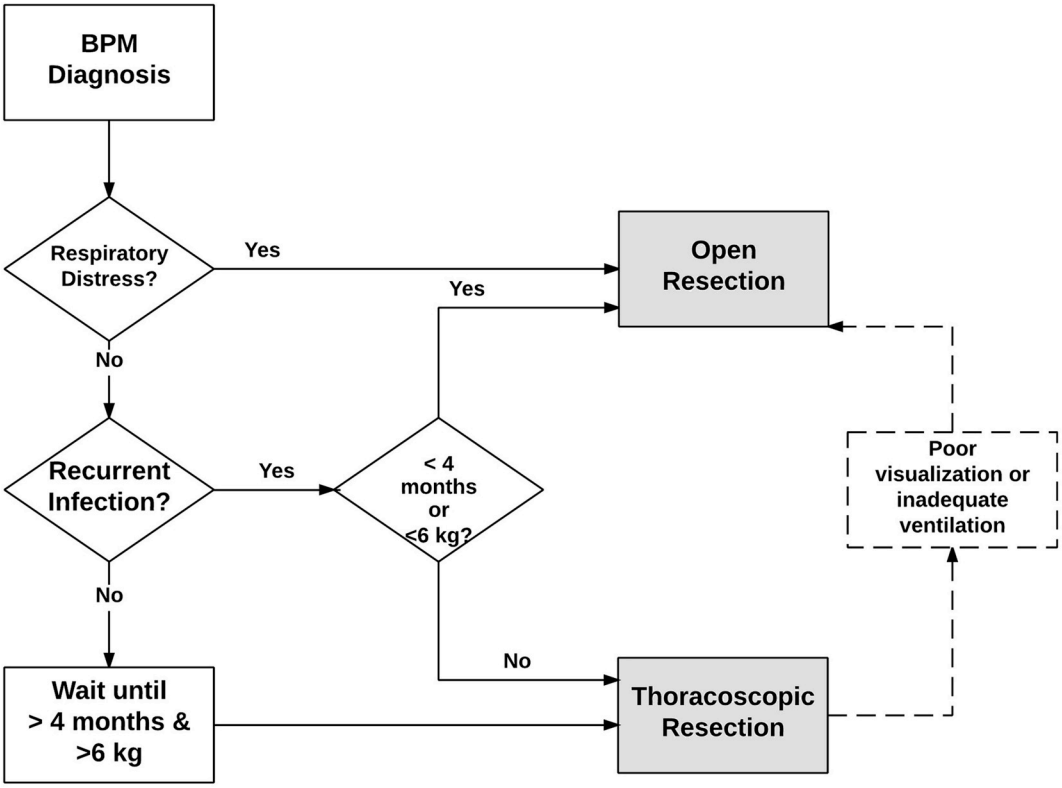

Fig. 6. Bronchopulmonary malformation surgical management algorithm utilized at Holtz Children's Hospital. Used with permissions.

the presence of EA. Barium contrast should be avoided as it can cause pneumonitis and granulomas if aspirated into the lungs. Patients with H-type TEFs may present early if the defect is large, with coughing and choking associated with feeding, but smaller defects of this type may not be symptomatic during the newborn period [25]. The diagnosis of H-type TEF (without EA) may not be evident on fluoroscopy and may require esophagoscopy and/or bronchoscopy to identify the fistula. Approximately one-half of cases of TEF and EA are associated with other anomalies, often as part of the VACTERL association (vertebral defects, anorectal malformation, cardiac defects, TEF, renal anomalies, and limb abnormalities), thus requiring further evaluation for additional anomalies [26].

The initial management for EA and TEF includes elevating the head to prevent aspiration of secretions, and decompression of the proximal esophageal pouch with a Replogle sump catheter. Routine endotracheal intubation should be avoided to prevent increased gastric distention from positive pressure ventilation, which can worsen pulmonary ventilation and even lead to gastrointestinal perforation. Preoperative echocardiography is necessary to rule out an anomalous right-sided aortic arch, as this finding may require a modified surgical approach. Early surgical intervention within the first few days of life is preferred for stable newborns of adequate birth weight.

The surgical treatment of TEF consists of ligation and division of the fistula. In cases of EA, primary anastomosis of the esophageal segments is preferred. A minimally invasive thoracoscopic approach is increasingly being utilized to minimize the morbidity associated with the traditional open thoracotomy [27]. Rarely, the distance between esophageal segments is too large for a primary anastomosis to be feasible, and surgical repair must be delayed for several months to allow for adequate esophageal growth to permit a primary repair. If this is unsuccessful, alternatives include elongation of the esophagus, interposition of jejunum or colon, or gastric transposition [28]. In preterm or very low birth weight infants (< 1500 g), surgical intervention within the first few days of life may be associated with increased morbidity, leading some pediatric surgeons to advocate for a staged approach with early ligation and division of the TEF followed by subsequent esophageal reconstruction at a later time [29,30]. In the setting of a delayed repair, early enteral access must be established for nutritional support. Feeding gastrostomy is rarely needed if early primary anastomosis is successful.

5. Congenital diaphragmatic hernia

Congenital diaphragmatic hernia (CDH) is a developmental defect of the diaphragm that allows abdominal viscera to herniate into the thoracic cavity. Due to direct compression of the ipsilateral lung and mediastinal shift and compression of the contra-lateral lung during development, there can be varying degrees of pulmonary hypoplasia and consequently, pulmonary arterial hypertension in the newborn. The vast majority of diaphragmatic defects are located posterior and lateral (Bochdalek hernia), with the remainder being anterior (Morgagni hernia), and rarely central. More than 80% of cases occur on the left side, and very rarely can be bilateral [31]. CDH is identified, or at least suspected, on routine prenatal ultrasound in more than half of cases. Affected neonates usually present in the first few hours of life with respiratory distress, although mild cases may not be identified until later in life. Physical findings include decreased or absent breath sounds on the affected side, a barrel-shaped chest, and a scaphoid-appearing abdomen due to migration of the abdominal contents into the thoracic cavity. On plain radiographs, the diagnosis of CDH can be quickly differentiated from large cystic anomalies of the lung by visualizing the course of a nasogastric tube that passes below the diaphragm with the tip curving back upward into the hemithorax within the herniated stomach.

When the diagnosis of CDH is made prenatally, or as soon as CDH is suspected after birth, patients who present with respiratory distress should be intubated immediately in the delivery room [32]. Bag-mask ventilation should be avoided to reduce gastrointestinal distention and further compression of the lung. The infant should be ventilated gently with low peak inspiratory pressures, employing permissive hypercapnia

to minimize lung injury [32,33]. Delay in securing an adequate airway may contribute to acidosis and hypoxia, which increase the risk of pulmonary hypertension. A nasogastric sump tube should be placed in the stomach and connected to continuous suction to decompress the abdominal contents and reduce lung compression. The administration of surfactant therapy does not appear to improve outcomes in infants with CDH [34]. Similarly, inhaled nitric oxide does not appear to have long-term benefits, despite its widespread use for treating pulmonary hypertension in patients with CDH [33,34]. Other vasodilatory agents (e.g. sildenafil, treprostinil) have been shown to confer some improvement in severe pulmonary hypertension in a few case series [33]. Echocardiography should be performed early to determine the severity of pulmonary hypertension and detect any associated cardiac anomalies, as significant heart defects are frequently associated with CDH and confer a worse prognosis.

ECMO is considered for infants who remain severely hypoxemic despite maximal conventional ventilatory support, as these patients will not survive without it. Indications for ECMO include persistent hypoxia despite maximal ventilator support, hypotension refractory to fluid resuscitation and inotropic support, and inadequate oxygen delivery with persistent metabolic acidosis. There is no difference in survival between veno-venous (VV) and veno-arterial (VA) ECMO, although VV ECMO is associated with fewer neurologic complications [33,35]. Infants on ECMO are at risk for intracranial bleeding due to the need for continuous anticoagulation. Therefore, frequent transcranial ultrasound examinations are recommended for the duration of therapy given that ECMO is contraindicated in the setting of severe intracranial hemorrhage. Although ECMO allows for preoperative stabilization, its continuation may be required for weeks until the pulmonary hypertension improves. In a small group of patients with severe pulmonary hypoplasia and/or pulmonary hypertension, there may be no response to therapy of any kind, including ECMO, and support is often withdrawn.

The timing of surgical repair of CDH is dependent upon first stabilizing the neonate. In patients with only mild symptoms on minimal support without evidence of pulmonary hypertension or hypoplasia, repair can typically be performed safely at 48–72 h after birth. In those with mild pulmonary hypoplasia or reversible pulmonary hypertension, surgery is delayed until pulmonary function improves and pulmonary hypertension is resolved, allowing CDH repair after 5–10 days. Controversy exists over the timing of operative repair in patients who require ECMO, with some advocating repair of the defect while the infant is on ECMO and others recommending repair only once the infant has been weaned off of ECMO. Regardless of timing, the surgical approach consists of reducing the abdominal viscera and closure of the diaphragmatic defect. Ideally, the defect can be closed with a primary suture repair, but a prosthetic patch repair is often required in patients with large defects in whom increased tension using a primary repair compromises thoracic compliance [36]. If the abdominal wall is difficult to close following reduction of the hernia, the use of a temporary abdominal wall silo or patch may be needed with delayed abdominal wall closure [37].

6. Poland syndrome

Poland syndrome is a rare constellation of congenital limb anomalies consisting of chest wall and upper extremity abnormalities that typically affect only one side. It is characterized by the partial or complete absence of the costal cartilages of multiple ribs, chest wall muscles, breast tissue and nipple, as well as a high-riding scapula and digital abnormalities (e.g. brachydactyly, syndactyly) [38]. Although most patients do not have respiratory symptoms, those with severe chest wall deformities may experience respiratory failure requiring mechanical ventilator support. These infants may exhibit paradoxical respiratory movement of the chest wall, similar to that seen in flail chest, due to multiple missing or incomplete ribs (Fig. 7A and B). In such cases, early surgical intervention is necessary to stabilize the chest wall using polytetrafluoroethylene or titanium mesh (Fig. 7C) to bridge the gap between the hypoplastic ribs and sternum. As these children grow, they will require additional reconstructive surgery and further thoracoplasty.

7. Abdominal wall defects

7.1. Gastroschisis

Gastroschisis is a full-thickness abdominal wall defect associated with evisceration of the bowel and is typically located to the right of the umbilical cord insertion. Most cases can be detected by prenatal ultrasound by the end of the first trimester, and nearly all cases are associated with an elevated maternal serum alpha-fetoprotein level [39]. Given that cesarean delivery has not been shown to improve neonatal outcomes for uncomplicated gastroschisis [40], the mode of delivery should be selected based on the usual obstetric indications.

The initial management after birth consists of wrapping the exposed bowel in sterile saline dressings and covering it with a plastic organ bag to minimize ongoing fluid and heat loss. Intravenous fluids and broad-spectrum antibiotics should be administered, and an orogastric tube inserted to decompress the stomach. Most affected infants are born prematurely; therefore, special attention must be given to the neonate's respiratory status and thermoregulation.

Surgical management consists of reducing the bowel into the abdomen and primary closure of the abdominal wall defect as soon as possible. Although this approach is feasible within a few hours of birth for many patients [41], some require delayed closure when the bowel is unable to be fully reduced. The bowel often develops inflammatory changes due to the prolonged exposure to amniotic fluid, resulting in thickened, distended intestinal loops. A silastic silo is applied for these cases, and the eviscerated bowel is gradually reduced over several days until operative fascial or sutureless flap closure can be achieved. Patients are maintained on total parenteral nutrition (TPN) until they gain bowel function, as prolonged postoperative dysmotility is common.

The majority of cases occur in isolation without other associated congenital anomalies or aneuploidy, thus conferring favorable prognosis with survival rates greater than 90% [40]. However, gastroschisis can be complicated by the presence of intestinal atresia, stenosis, perforation, necrosis, or volvulus. The occurrence of these complications may be related to vascular disruption caused by herniated bowel and is associated with increased morbidity and mortality [42]. Nonetheless, the prognosis for infants born with gastroschisis generally remains favorable even in these complicated cases.

7.2. Omphalocele

Omphalocele is a midline abdominal wall defect located at the base of the umbilical cord. The defect is covered by a membranous sac that contains the herniated abdominal contents, and the umbilical cord inserts at the apex of the sac. When the defect is less than 4 cm in size, it is simply referred to as a “hernia of the umbilical cord.” Giant omphalocele refers to larger defects (> 5 cm) that also contain most of the liver. Omphalocele is usually detected by fetal ultrasound during the first or second trimester, with over 90% of cases diagnosed prenatally. A high frequency of fetal aneuploidy, particularly trisomy 18 or 13, is associated with omphalocele [43].

Similar to gastroschisis, the initial management of omphalocele begins with preservation of the sac with sterile saline-soaked gauze or a plastic organ bag, gastric decompression with an orogastric tube, and the administration of intravenous fluids and antibiotics. During the delivery, care must be taken to avoid clamping the umbilical sac, especially in cases of umbilical cord hernia where the small sac can be mistaken for the proximal part of the umbilical cord. Given that approximately half of affected infants have other associated anomalies, a thorough diagnostic workup must be performed to exclude sternal cleft,

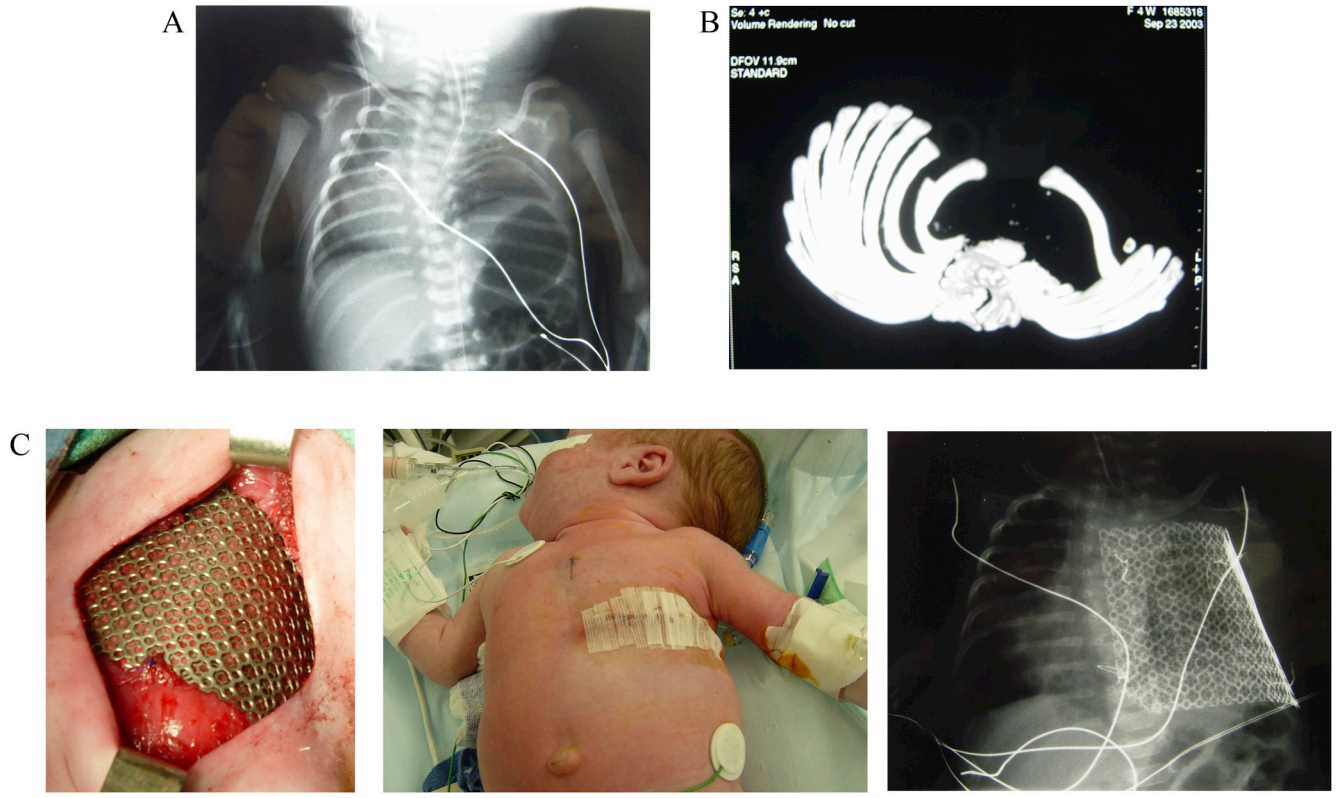

Fig. 7. Premature infant with severe chest wall deformity due to Poland syndrome chest radiograph (A), computed tomography reconstruction (B), and surgical repair using titanium mesh (C). Used with permissions.

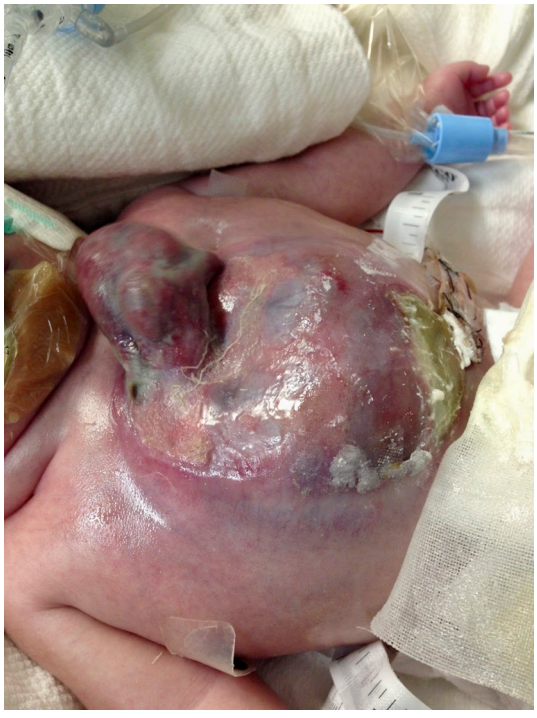

Fig. 8. Premature infant with Pentalogy of Cantrell including omphalocele and ectopia cordis.

ectopia cordis, cardiac defects, diaphragmatic hernia, bladder exstrophy, and cloacal anomalies (Fig. 8). With regard to surgical management, small defects can usually be closed primarily within 24–72 h after birth. Larger defects may require delayed closure following the sequential reduction of the herniated organs using a silastic silo over three to seven days. In cases of giant omphalocele, repair of the defect can be achieved by a combination of silastic silo reduction, prosthetic patch closure, and skin grafting or application of topical sclerosing agents (i.e. silver sulfadiazine) to expedite eschar formation with a delayed hernia repair [44].

8. Conclusions

In summary, critical congenital malformations of the newborn require early detection and prompt intervention to minimize associated morbidity and mortality. In the absence of prenatal detection, the majority of these anomalies present with respiratory distress or obvious external malformations in the early neonatal period. Clinicians must maintain a high index of suspicion for these relatively rare conditions in an effort to quickly secure the correct diagnosis. Ideally, consultation with the pediatric surgeon should occur well before the end of pregnancy when these critical malformations are identified prenatally.

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Declaration of competing interest

None declared.

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