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Isolated Thoracoschisis with Rib Agenesis and Liver Herniation: A Case Report

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Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
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Patient: Male, 30-day-old
Final Diagnosis: Thoracoschisis
Symptoms: Respiratory distress
Medication: —
Clinical Procedure: —
Specialty: Surgery

Objective: Congenital defects/diseases

Background: Thoracoschisis is a very rare congenital birth defect defined by the herniation of intra-abdominal organs through a defect in the thoracic wall. Though often associated with other birth defects as a part of the "limb-body wall complex" deformities, thoracoschisis has very rarely been reported as an isolated finding.

Case Report: Here we present the case of a 30-day-old term male infant with an isolated left thoracoschisis managed successfully by primary closure. The patient was monitored postnatally in the Neonatal Intensive Care Unit (NICU) of Maputo Central Hospital because of the presence of a herniated mass through a left chest wall defect below the left nipple. Computed tomography (CT) scans suggested the presence of a left diaphragmatic hernia, left rib agenesis, and herniation of an unidentifiable intra-abdominal organ through the anterior left chest wall. On day of life (DOL) 30, when global health outreach pediatric surgeons arrived at the hospital, the decision was made to operate on the child. The mass was found to be of liver origin, the exposed tissue was excised, and primary closure of the chest wall was accomplished. The patient's postoperative course involved a wound infection that resolved favorably with treatment, allowing for discharged home on postoperative day (POD) 17 in stable condition.

Conclusions: Our case report highlights the importance of recognizing this rare condition and directing appropriate surgical care.

MeSH Keywords: Congenital Abnormalities • Congenital, Hereditary, and Neonatal Diseases and Abnormalities • Hernia • Mass Chest X-Ray • Pediatrics

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Background

Thoracoschisis is a rare congenital birth abnormality consisting of the herniation of intra-peritoneal organs through a defect in the thoracic wall. Thoracoschisis is classified as a part of the "Limb-Body Wall Complex" (LBWC) deformities, being more commonly associated with other birth defects such as limb agenesis, anterior thoracic wall deformities, phocomelia, and diaphragmatic hernias [1–6]. However, of the 14 total reported cases of thoracoschisis, 5 cases had no other related abnormalities [7–11].

The etiology behind thoracoschisis is still unidentified, but the presence of the other associated LBWC deformities gives insight into the timing at which these deformities likely develop [10]. Initial theories behind the pathogenesis of LBWC deformities include abnormal embryonic folding, formation of amniotic bands that interfere with limb development, rupture of amniotic sacs, and vascular malformations [3,12,13].

Diagnosis of the full extent of the defect differs on a case-by-case basis, but usually involves chest radiography (CXR) or computed tomography (CT) scans [4]. Exploratory operations are often necessary as it is sometimes unclear whether there is an elevated hemidiaphragm or a diaphragmatic hernia [8,10]. Prenatal diagnosis of thoracoschisis has been successfully demonstrated using 2D and 3D ultrasound [14]. Treatment of thoracoschisis is varied but involves primary management to assure hemodynamic and respiratory stability, followed by imaging for diagnosis, and finally either surgical reduction of abdominal contents through subcostal incision or gastroschisis-like silo bags. This is followed by closure of the thoracic wall, re-approximation of separated ribs, or plans to do staged reconstruction of the thoracic wall post-operatively [10,11]. In this case report we present a rare instance of a term male infant with an isolated left-sided thoracoschisis and rib agenesis.

Case Report

During a one-week global health outreach to Maputo, Mozambique, a 30-day old male infant was identified in the NICU of Maputo Central Hospital, Mozambique with a 3×3 cm mass overlying the left chest wall located below the left nipple (Figure 1). The mass was nonreducible, contiguous with the surrounding skin, and covered by friable granulation tissue. Upon further examination of the chest wall, there was a large surrounding defect in the rib cage around the protruding mass. There were no underlying palpable ribs at the location of the mass and no pectoral muscle present on the affected side. In addition, there was an accessory nipple complex located inferior to the mass. The rest of the physical examination was unremarkable.

Figure 1. Showing the 3×3 cm herniation of the left lobe of the liver overlying the left chest wall.

Upon review of the patient's medical records we discovered that the infant was born by means of vaginal delivery to an otherwise healthy 20-year-old female in Tete province, Mozambique. Of note, she had received no prenatal care and contracted malaria during the first trimester of pregnancy. Immediately after birth the child was stable, needing no ventilatory assistance. He was subsequently transferred to Maputo Central Hospital NICU for further management of the left chest wall mass. The patient was evaluated by an echocardiography, which was deemed normal, as well as CXR and a CT of his chest (Figure 2), which delineated a possible left-sided diaphragmatic hernia, with multiple partial rib agenesis surrounding the area of the chest wall herniation. It was unclear based on the CT images whether the herniated mass contained parts of the left liver or lung.

The decision was made to take the child to the operating room (OR) for definitive repair. As the initial images were suggestive of an associated left diaphragmatic hernia, we approached the repair by left subcostal incision (Figure 3). It became immediately obvious that the diaphragm was very high in the left chest, but was intact and without a defect. Next, we tried to reduce the mass back into the abdominal cavity, but without success. Mobilization of the mass from the chest wall was accomplished by a combination of blunt and electrocautery dissection. Upon further inspection, we determined that the

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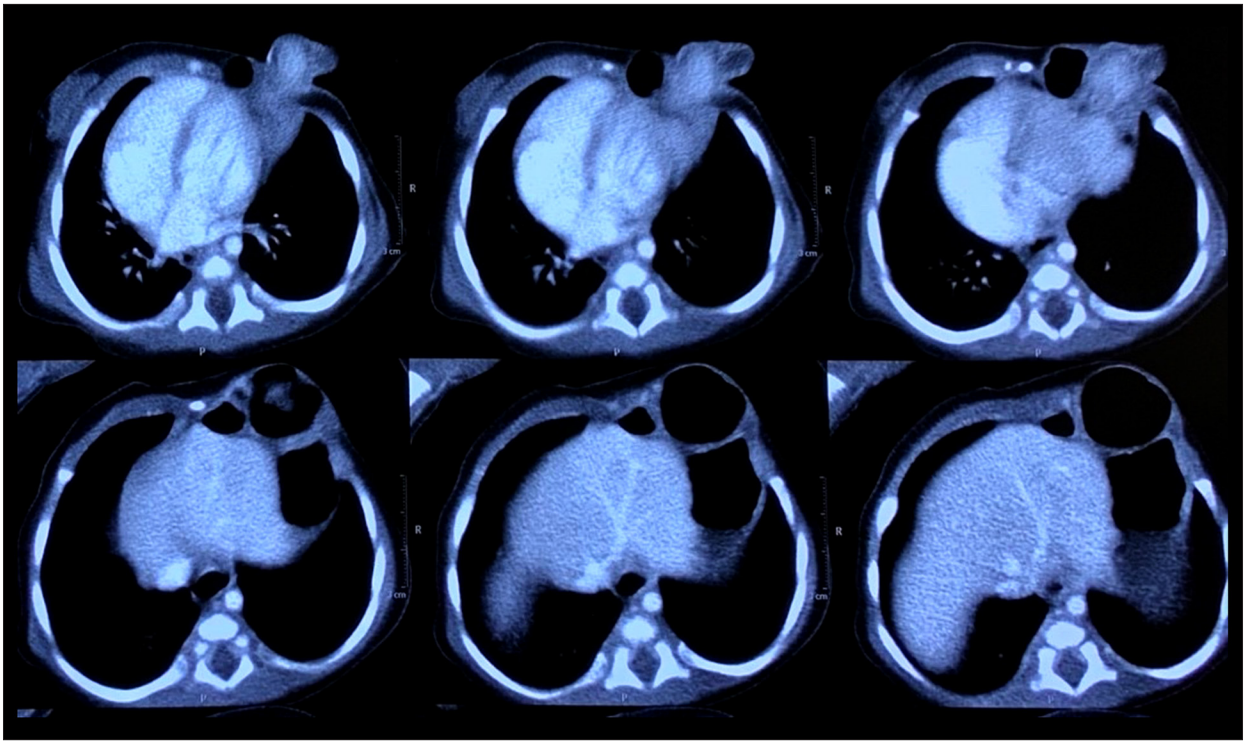

Figure 2. Axial CT images demonstrating herniation and rib agenesis.

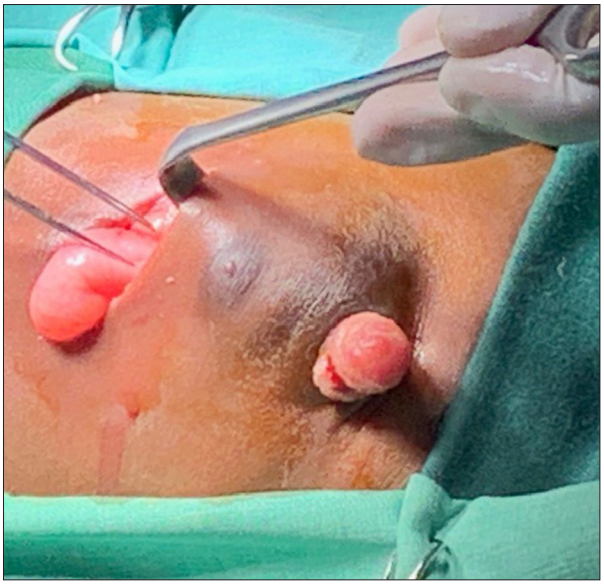

Figure 3. A subcostal incision was made to address the presumed left-sided diaphragmatic hernia.

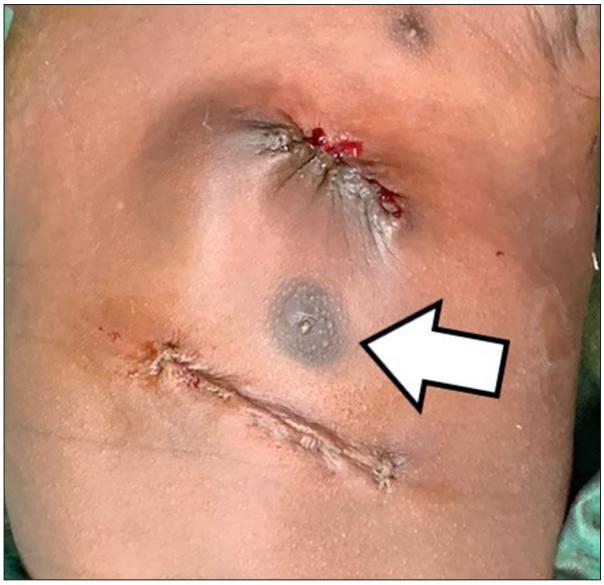

Figure 4. After reduction and closure of the thoracic wall defect, as well as the subcostal incision. * Note the presence of an accessory nipple complex located inferior to the chest wall defect.

mass was originating from the left lobe of the liver. At this point, we decided to excise the exposed tissue at the level of the chest wall and allow the liver to retract back into the abdomen. The chest wall was then closed in layers (Figure 4). Postoperative recovery was complicated by the presence of an infection at the wound site, which subsequently resolved with antibiotic treatment, allowing for discharge on POD 17.

Discussion

Thoracoschisis is a very rare congenital abnormality, having only been reported 14 other times in the medical literature (Table 1). In direct contrast to thoracoabdominoschisis,

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Table 1. Thoracoschisis: a review of literature.

Reference	Year	Gender	Defect location	Content	Associated malformation	Survival
Davies et al. [1]	1977	Female	Left third intercostal	Left liver lobe, stomach, transverse colon	No left forearm, syndactyly, dextrocardia	Alive
Bamforth et al. [15]	1992	Female	Left sixth rib	Left liver lobe	Left Poland anomaly, scapula hypoplastic, no humerus, no ulna, no radius, dextrocardia	Alive
Derbent et al. [6]	2001	Female	Right second to fourth rib	Liver, intestine	LBWC, other deformations not able to be defined	Intrauterine Death
Biri et al. [2]	2006	Female	Left	Left liver lobe	Left forearm agenesis, right arm-hand agenesis	Died at Birth
Karaman et al. [7]	2011	Male	Left eighth intercostal	Liver, transverse colon, omentum	None	Alive
Bhattacharyya et al. [5]	2012	Female	Right	Riedel liver lobe, stomach, small intestine	Agenesis of the right upper limb, right upper quadrant abdominal wall defect	Died at Birth
Eck et al. [16]	2015	Male	Right fifth–eight ribs	Liver, intestine, omentum	Right fourth digit abnormality	Alive
McKay et al. [17]	2015	Female	Left eighth intercostal	Riedel liver lobe, omentum	Positional deformity of left hand & palmar contractures of the fingers	Alive
Seleim et al. [8]	2015	Male	Left 4th intercostal	Riedel liver lobe, stomach, intestine	None	Died 2 days post operative
Travers et al. [9]	2016	Female	Left rib aplasia	Mesenchymal hamartoma	None	Alive
de Grijs et al. [10]	2017	Female	Left of fourth–sixth ribs	Liver, stomach, transverse colon	None	Alive
Vujovic et al. [4]	2017	Female	Right first intercostal	Riedel liver lobe	Hypoplasia of the right arm and incomplete hand duplication	Alive
Hanafi et al. [14]	2017	Male	Left third and fourth ribs	Left liver lobe, stomach, spleen, intestine	Radial aplasia, unilateral polydactyly and syndactyly	Died at DOL 3
Harris et al. [11]	2017	Male	Left Ribs	Liver	None	Alive

thoracoschisis is not a midline defect and does not involve the abdominal wall. Davies et al. first described it in 1977 as being associated with chest wall malformations and diaphragmatic hernias [1]. Since then, other cases have been noted to be associated with limb abnormalities, being classified as a part of the group of defects known as "Limb-Body Wall Complex" (LBWC) deformities [3]. Only 5 of the reported cases of thoracoschisis have had no other related defects, termed "isolated thoracoschisis". Our case falls into the category of isolated thoracoschisis.

In a similar case report of isolated thoracoschisis, Saleim et al. noted an elevated left hemi-diaphragm, with a 3×2 cm thoracic wall defect in the 4th intercostals space. The herniated contents of the thoracoschisis were the liver, stomach, and small bowel. Because the small bowel was involved, a gastroschisis-like silo bag was created to assist in reduction [8].

In some cases, as described by Karaman et al. and Grijs et al., the herniated tissue, consisting of either liver, stomach, omentum, or transverse colon, was able to be separated from adhesions to the chest wall, so a simple reduction and primary repair was successful [7,10]. Our case falls more into the group of cases described by Travers et al. and Harris et al., in which the external herniated tissue, mainly liver, needed to be resected prior to reduction [9,11]. These 2 reports are similar

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to our case, as the only tissue herniated through the thoracic wall defect was part of the liver.

An important distinction between different cases of isolated thoracoschisis is the presence of rib agenesis. In reports by Harris et al., Saleim et al., and Karaman et al., there was an enlarged intercostal space between the ribs, but no rib agenesis was present [7,8,11]. In comparison, Grijs et al. and Travers et al. noted segmental agenesis of the involved ribs [9,10]. To the best of our knowledge, the present case is the third reported case of isolated thoracoschisis with rib agenesis.

Current survival rates of all forms of thoracoschisis are about 66%, and are mainly dependent on the associated malformations (Table 1). Of the 5 reported cases of thoracoschisis that resulted in death, 1 was intrauterine with undiagnosed associated malformation, 3 were associated with limb malformations, and 1 was isolated [2,5,6,8,14]. For those that survived reduction of the herniated contents, no post-operational complications were noted. Those with rib cage and limb malformations were scheduled for reconstructive surgery.

Conclusions

Isolated thoracoschisis with rib agenesis is a very rare congenital abnormality, which, to the best of our knowledge, has only being described 2 other times in the medical literature. Our case demonstrates a less severe form of isolated thoracoschisis that was successfully managed by primary surgical repair.

Conflict of interest

None.

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