Isolated thoracoschisis: Case report

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Thoracoschisis is a rare congenital anomaly that refers to a congenital fissure of the chest wall. It is frequently accompanied with other congenital defects of the limbs and the abdominal wall as part of the limb-body wall complex, which is exencephaly/encephalocele and facial clefts, thoracoschisis and/or abdominoschisis and limb defects. Isolated thoracoschisis is a rare entity. We present a case of isolated thoracoschisis. A 24-week gestational age boy presented with a 3 cm chest wall defect in the left lateral 10th intercostal space and intestines herniating through the defect. There was no history of maternal drug use during pregnancy. Birth weight was 500 g. He underwent surgery. The intestines were reduced via the thoracic wall defect.

Key words: congenital defect, limb-body wall complex, thoracoschisis.

Thoracoschisis is a rare congenital anomaly characterized by evisceration of intra-abdominal organs through a thoracic wall defect1. Thoracoschisis resembles gastroschisis which is characterized by a defect on the anterior abdominal wall. However, the defect is on thoracic wall in the thoracoschisis. The etiology of this congenital anomaly is unknown. Thoracoschisis is frequently accompanied with other congenital anomalies of the limbs and the abdominal wall as part of the limb-body wall complex (LBWC) which is exencephaly/encephalocele and facial clefts, thoracoschisis and/or abdominoschisis and limb defects2,3. Isolated thoracoschisis is a very rare condition. To the best of our knowledge; this is the 4th case of isolated thoracoschisis reported in the English medical literature, which does not include evisceration of the liver.

Case Report

The patient was a male premature infant, with a gestational age of 24-weeks, born to a 40-year-old mother by urgent Cesarean section due to preterm delivery. There was first cousin consanguinity between parents. There was no history of maternal drug use during pregnancy. Birth weight was 500 g and Apgar scores were 4 and 7 in 1st and 5th minutes, respectively. Prenatal follow-up was inadequate with no ultrasonographic examination. The patient was intubated, stabilized and then received surfactant (Curosurf®, 200 mg/kg) in the delivery room and was admitted to the neonatal intensive care unit. Physical examination revealed a 3 cm chest wall defect in the left lateral 10th intercostal space and small intestines and colon eviscerating through the defect (Fig. 1). Transverse colon seemed ischemic. Intestines were wrapped with sterile wet hot gauze. Abdomen was scaphoid. Upper and lower extremities were normal and there were no other dysmorphic features. Postnatal echocardiography revealed patent ductus arteriosus and patent foramen ovale. Abdominal and chest X-ray demonstrated bilateral normal diaphragm, gastric air on left upper quadrant and gas filled loops of intestine protruding from lateral side of left hemi thorax (Fig. 2). The patient underwent surgery on the third hour of his life. The intestines were reduced via the thoracic wall defect under general anesthesia. Abdominal cavity was explored through the defect and an additional incision was not necessary. Left hemi diaphragm was intact and there was no abdominal visceral anomaly. After reduction of eviscerated intestines, transverse colon seemed...
<table>
<thead>
<tr>
<th>Author</th>
<th>Sex</th>
<th>Defect location</th>
<th>Herniated organs</th>
<th>Diaphragmatic defect / abnormality</th>
<th>Additional abnormality</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Davies et al.¹</td>
<td>Female</td>
<td>Left third intercostal</td>
<td>Liver left lobe, stomach, transverse colon, omentum</td>
<td>Left anterolateral</td>
<td>LBWC (No left forearm), syndactyly, dextrocardia</td>
<td>Alive</td>
</tr>
<tr>
<td>Bamforth et al.¹¹</td>
<td>Female</td>
<td>Left sixth rib</td>
<td>Liver left lobe</td>
<td>Left posterior</td>
<td>LBWC (left Poland anomaly, left scapula hypoplastic, no humerus, no ulna, no radius), dextrocardia, ectopic pancreas in the duodenum, intestines in the left hemithorax</td>
<td>Alive</td>
</tr>
<tr>
<td>Derbent and Balci²</td>
<td>Female</td>
<td>Right second to fourth ribs</td>
<td>Liver, intestines</td>
<td>Right anterolateral</td>
<td>LBWC</td>
<td>Intrauterine death</td>
</tr>
<tr>
<td>Biri et al.¹²</td>
<td>Female</td>
<td>Left / unspecified thoracic location</td>
<td>Liver left lobe</td>
<td>Hiatus hernia</td>
<td>LBWC (left forearm agenesis), ventricular septal defect</td>
<td>Died at birth</td>
</tr>
<tr>
<td>Karaman et al.¹³</td>
<td>Male</td>
<td>Left eighth intercostal</td>
<td>Liver, transverse colon, omentum</td>
<td>Left diaphragm evagination</td>
<td>Atrial septal defect, patent ductus arteriosus</td>
<td>Alive</td>
</tr>
<tr>
<td>Bhattachryya et al.¹⁴</td>
<td>Female</td>
<td>Right unspecified thoracic location</td>
<td>Liver, colon, small intestines, spleen</td>
<td>Absence of right dome of diaphragm, herniation</td>
<td>LBWC, right pulmonary hypoplasia, heart and lung exposed to air, right upper limb agenesis, absence of right ribs</td>
<td>Died at birth</td>
</tr>
<tr>
<td>Seleim et al.¹⁵</td>
<td>Male</td>
<td>Left fourth intercostal</td>
<td>Riedel liver lobe, stomach, intestines</td>
<td>Left diaphragm evagination</td>
<td>Dextrocardia, patent ductus arteriosus</td>
<td>Died after surgery</td>
</tr>
<tr>
<td>McKay et al.¹⁶</td>
<td>Female</td>
<td>Left seventh intercostal</td>
<td>Riedel liver lobe, omentum</td>
<td>Left Morgagni hernia</td>
<td>Septal defects, patent ductus arteriosus, left hand palmar contractures</td>
<td>Alive</td>
</tr>
<tr>
<td>Our case</td>
<td>Male</td>
<td>Left tenth intercostal</td>
<td>Transverse colon, small intestine</td>
<td>No</td>
<td>Patent ductus arteriosus, patent foramen ovale</td>
<td>Died after surgery</td>
</tr>
</tbody>
</table>

LBWC: limb body wall complex, PDA: patent ductus arteriosus
better and there was no need for resection (Fig. 2). Postoperative X-ray demonstrated bilaterally normal hemi-diaphragms and normal intestinal gas distribution (Fig. 3). The patient died on the third postoperative day due to multi-organ failure. Autopsy and genetic evaluation was not possible since the parent did not give consent. The parent allowed the images and information to be used in the article.

Discussion

Thoracoschisis is a congenital anomaly, which is frequently accompanied with congenital defects of the limbs and the abdominal wall as part of the limb-body wall complex. The diagnostic criteria for LBWC is presence of at least two of the following three malformations; exencephaly/encephalocele and facial clefts, thoracoschisis and/or abdominoschisis and limb defects.

Isolated thoracoschisis is a much rare entity defined with evisceration of intraabdominal organs through thoracic wall. Most of the isolated thoracoschisis cases reported have ipsilateral diaphragmatic hernia/eventration and all include evisceration of liver. Left sided thoracoschisis is most common and it is also predominant in females (Table 1). Our case is the first reported case of isolated thoracoschisis, which does not involve eviscerated liver, diaphragmatic abnormality and LBWC. Considering reported cases of LBWC and isolated thoracoschisis including a wide range of congenital defects, this rare clinical entity should be defined as a heterogeneous
group of anomalies with different phenotypic occurrences. Previous classifications do not cover all cases reported 4-7.

The etiology of this congenital anomaly is unknown. Theories on the pathogenesis of the LBWC are germ disc defect with early embryonic maldevelopment, primary rupture of the amnion leading to the formation of amniotic bands, vascular disruption and disturbance of the embryonic folding process 1-3, 8,9. Etiology has not been clearly associated with teratogenic agents and genetic abnormalities 10.

Antenatal diagnosis is usually based on ultrasound examination. Prenatal magnetic resonance imaging demonstrates anatomic details defining the anomaly. Early diagnosis will allow appropriate management including termination of pregnancy according to the severity of congenital defects included. Eviscerated bowels should be wrapped with sterile wet gauze or sterile plastic bag in the delivery room to prevent heat and fluid loss and protect intestinal injury.

Survival depends on the extend of anomaly as well as gestational age of the newborn. Other reported cases 1-7 in the literature without severe anomalies had chance of survival and survival seems to be higher in the cases with higher birth weight and older gestational age.

REFERENCES