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Isolated thoracoschisis: Case report

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SUMMARY: Ardıçlı B, Karaman A, Özyazıcı A, Zenciroğlu A, Okumuş N. Isolated thoracoschisis: Case report. Turk J Pediatr 2017; 59: 217-220.

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 defect[1]. 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 defects[2,3]. 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 bilaterally 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 not any abdominal visceral anomaly. After reduction of eviscerated intestines, transverse colon seemed

Table I. Thoracoschisis Cases

Author	Sex	Defect location	Herniated organs	Diaphragmatic defect / abnormality	Additional abnormality	Result
Davies et al.[1]	Female	Left third intercostal	Liver left lobe, stomach, transverse colon, omentum	Left anterolateral	LBWC (No left forearm), syndactly, dextrocardia	Alive
Bamforth et al.[11]	Female	Left sixth rib	Liver left lobe	Left posterior	LBWC (left Poland anomaly, left scapula hypoplastic, no humerus, no ulna, no radius), dextrocardia, ectopic pancreas in the duodenum, intestines in the left hemithorax	Alive
Derbent and Balci[2]	Female	Right second to fourth ribs	Liver, intestines	Right anterolateral	LBWC	Intrauterine death
Biri et al.[12]	Female	Left / unspecified thoracic location	Liver left lobe	Hiatus hernia	LBWC (left forearm agenesis), ventricular septal defect	Died at birth
Karaman et al.[13]	Male	Left eighth intercostal	Liver, transverse colon, omentum	Left diaphragm eventration	Atrial septal defect, patent ductus arteriosus	Alive
Bhattachryya et al.[14]	Female	Right unspecified thoracic location	Liver, colon, small intestines, spleen	Absence of right dome of diaphragm, herniation	LBWC, right pulmonary hypoplasia, heart and lung exposed to air, right upper limb agenesis, absence of right ribs	Died at birth
Seleim et al.[15]	Male	Left fourth intercostal	Riedel liver lobe, stomach, intestines	Left diaphragm eventration	Dextrocardia, patent ductus arteriosus	Died after surgery
McKay et al.[16]	Female	Left seventh intercostal	Riedel liver lobe, omentum	Left Morgagni hernia	Septal defects, patent ductus arteriosus, left hand palmar contractures	Alive
Our case	Male	Left tenth intercostal	Transverse colon, small intestine	No	Patent ductus arteriosus, patent foramen ovale	Died after surgery
Total (n)	Female: 6 Male: 3	Left: 7 Right: 2	Liver: 8 Colon: 3 Small intestine: 4 Stomach: 2 Omentum: 2	Diaphragmatic defect / abnormality : 8	LBWC: 5 PDA: 4 Dextrocardia: 3 Septal defect: 3	Alive: 4 Fatal: 5

LBWC: limb body wall complex, PDA: patent ductus arteriosus

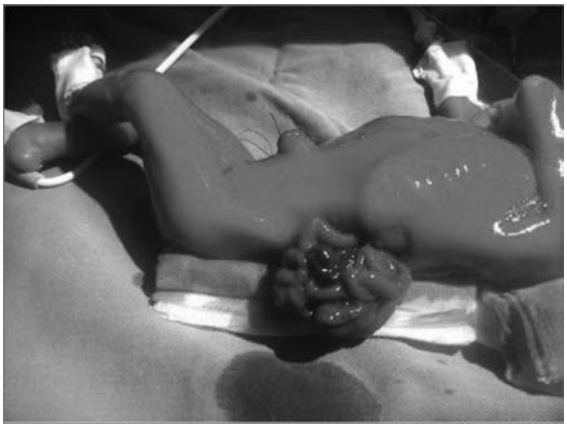

Fig. 1. The intestinal segments herniated through the thoracic defect. The circulation of the transverse colon was damaged.

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[3].

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

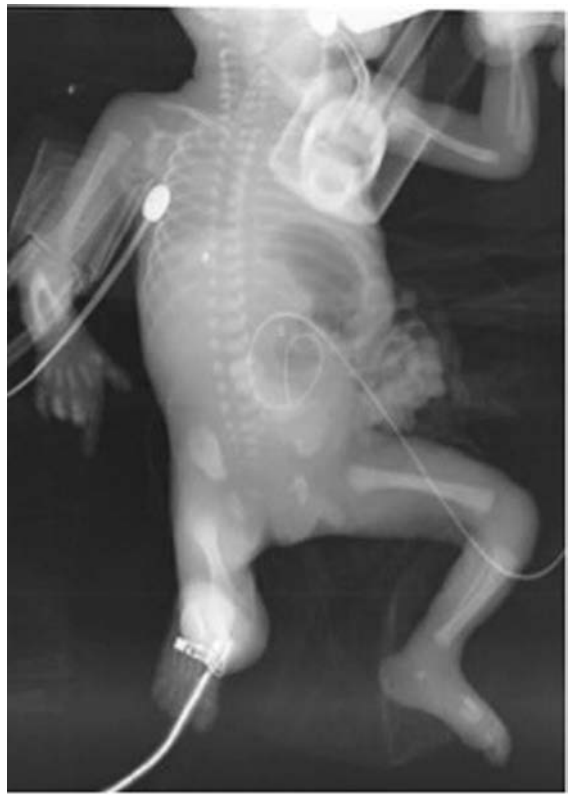

Fig. 2. Preoperative X-ray demonstrating normal diaphragm, gastric air on left upper quadrant and intestines protruding out.

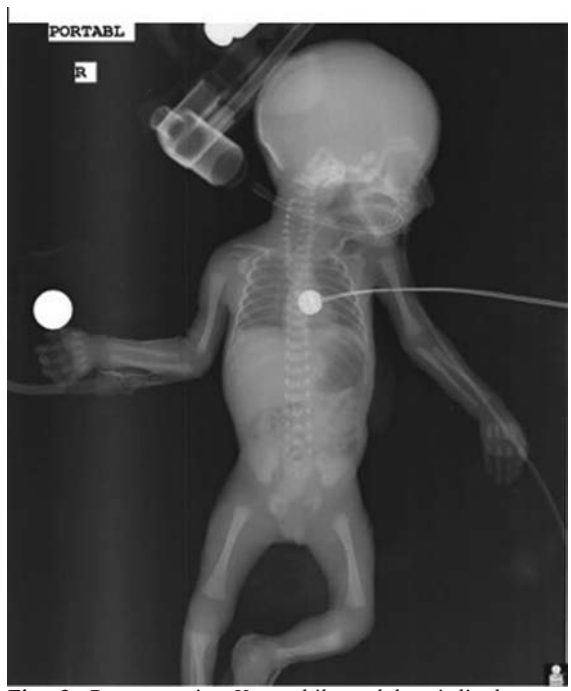

Fig. 3. Postoperative X-ray, bilateral hemi-diaphragms normal position and normal intestinal gas distribution.

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.

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