# A Rare Cause of Respiratory Distress in a Newborn: Mediastinal Enteric Cyst Associated with Costovertebral Malformation: Case Report

## Yenidoğanda Nadir Bir Solunum Sıkıntısı Nedeni: Kostovertebral Malformasyona Eşlik Eden Mediastinal Enterik Kist

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**ABSTRACT** Mediastinal enteric cyst is usually associated with segmental costovertebral malformations include Jarcho-Levin syndrome, spondylocostal dysostosis, and spondylothoracic dysostosis. We aimed to report a case of thoracal vertebral anomaly associated with a mediastinal enteric cyst. A 1 day-old girl patient was admitted to our clinic with symptoms of dyspnea, cyanosis, grunting and poor sucking. Low hairline, a short webbed neck, and flexion contracture in the left fourth phalanx were detected in her physical examination. Posteroanterior chest X-ray and thorax computerized tomography demonstrated a large cystic lesion at the right lung. Herein this newborn with respiratuar distress due to mediastinal enteric cyst associated with costovertebral malformation was reported.

**Key Words:** Dyspnea; infant, newborn; mediastinal cyst

**ÖZET** Mediastinal enterik kist genellikle segmental kostovertebral malformasyonlarla karakterize Jarcho-Levin sendromu, spondilokostal ve spondilotorasik disostoza eşlik eder. Bu yazıda, torakal vertebral anomaliye eşlik eden mediastinal enterik kist bulunan yenidoğan bir hastayı sunmayı amaçladık. Bir günlük yenidoğan kız hasta kliniğimize solunum sıkıntısı, morarma, hırıltılı solunum ve emme güçlüğü şikayetleri ile başvurdu. Fizik incelemede hastada düşük ense saç çizgisi, kısa boyun ve sol 4. el parmağında fleksiyon kontraktürü saptandı. Çekilen ön arka akciğer grafisi ve bilgisayarlı göğüs tomografisinde sağ akciğerde geniş bir kistik lezyon tespit edildi. Solunum sıkıntısına neden olan ve kostovertebral malformasyonun eşlik ettiği, mediastinal enterik kist saptanan yenidoğan bu olgu rapor edildi.

**Anahtar Kelimeler:** Solunum sıkıntısı, yenidoğan, mediastinal kist

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Foregut duplication cysts constitute 10% of all mediastinal tumours.[1] Three types of foregut cysts have been described: bronchogenic, intramural esophageal and enteric. The diagnosis basically depends on the histological findings. However, vertebral anomalies associated with posterior mediastinal cyst is a feature of enteric cyst. Enteric cyst is uncommon and none of the 15 cases of foregut cyst in children, which were reported by Cohen, et al.[2] was of enteric variety. The high incidence of associated thoracic or cervical vertebral anomalies with foregut cysts provide an early clue to the diagnosis. The enteric cyst has been generally reported to be at the right side of the mediastinum and asymptomatic. If

symptomatic, it may lead to respiratory insufficiency and pneumonia due to the compression of the pulmonary parenchyma. Also, signs of obstruction due to pressure on the esophagus have been reported.[3]

Segmental costovertebral malformations include Jarcho-Levin syndrome (JLS), spondylocostal dysostosis (SCD), and spondylothoracic dysostosis (STD). Spondylocostal dysplasia, which is also known as SCD, is a congenital segmental costovertebral malformation with multiple vertebrae and numerical or structural rib abnormalities resulting in thoracic asymmetry, short stature, and a short neck. On the other hand, STD, which involves the spine, leads to a fanlike chest but bears no intrinsic rib malformation. On the other hand, JLS is a severe form of the anomaly which involves the whole vertebral column.[4]

Here, a newborn with respiratuar distress due to mediastinal enteric cyst with costovertebral malformation has been reported.

## CASE REPORT

A 1 day-old girl was born at term, after via spontaneous vaginal delivery, from a 35 years old woman, as her seventh live birth. The baby was admitted to our clinic with symptoms of cyanosis, grunting and poor sucking. Her medical history and family history were unremarkable.

Physical examination revealed the following findings: body weight 3400 g (50th percentile); height 48 cm (25-50 percentile); head circumference 34.5 cm (50 percentile); respiratory rate 44/min; blood pressure 50/30 mmHg; pulse rate 148/min; oxygen saturation 78%; and body temperature 36.1°C. She was grunting, cyanotic at finger tips and lips. She had short webbed neck, low hairline (Figure 1), high arched palate, and flexion contracture of the left fourth phalanx. On auscultation, secretory crepitations were detected at both lung fields. There were intercostal and subcostal retractions.

Complete blood count revealed a hemoglobin level of 14 g/dL; leukocyte and platelet counts of 13.400/mm$^3$ and 206.000/mm$^3$, respectively. Blood

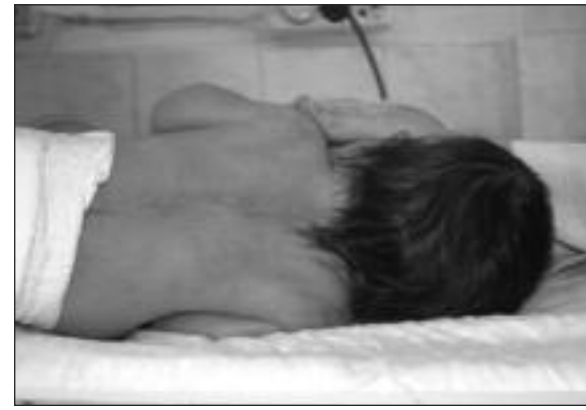

**FIGURE 1:** Low hairline and short neck.

gas analysis revealed the following findings: pH, 7.47; $pCO_2$, 57 mmHg; and $pO_2$, 45 mmHg. She was chromosomally a normal girl (46,XX).

Posteroanterior chest X-ray demonstrated extensive segmentation deformities on the thoracal vertebrae. There was a cystic lesion replacing two thirds of the right lung and fusion on the thoracal vertebrae (Figure 2a and 2b). Thorax CT examination revealed a round cystic lesion of 6x6 cm with smooth surfaces in the right lung (Figure 3). Echocardiography and abdominal US findings were normal.

Total cystectomy was performed. During the postoperative period, oxygen saturation was maintained between 92% and 96%. Biochemical analysis of the cystic material was as follows: glucose 15 mg/dL, LDH 162 U/L, and chloride 115 mg/dL. In the pathological examination, a cystic formation with a fibromuscular wall lined with intestinal epithelium having villous protrudings towards the surface was observed (Figure 4).

The patient was discharged on seventh postoperative day. Only minimal limitation in the neck movements was noted postoperatively, but did well otherwise. Her breath sounds was normal on auscultation, and her oxygen saturation was 96% in last clinical control.

## DISCUSSION

Mediastinal enteric cysts are rare clinical entities, which have been generally reported to be asymp-

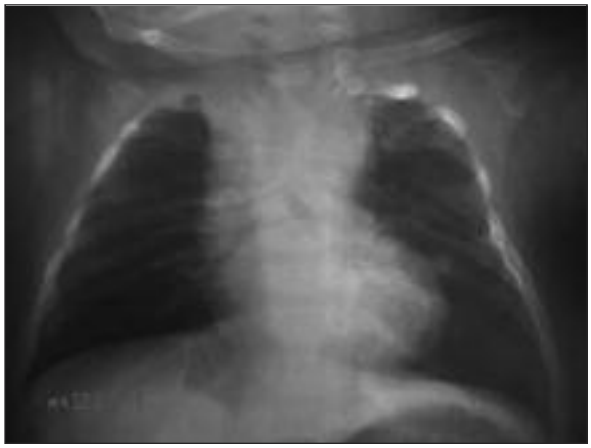

**FIGURE 2a:** Posterior-anterior chest X-ray. Preoperative appearance.

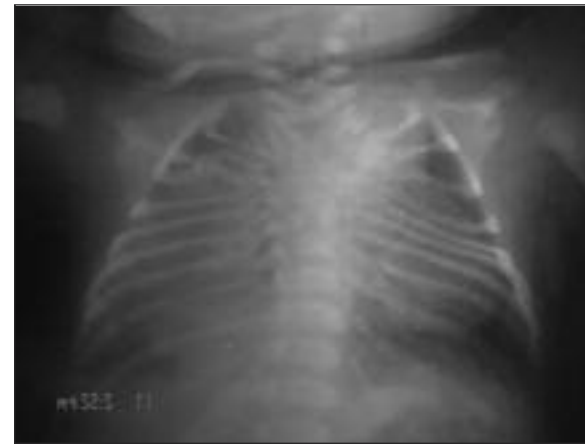

**FIGURE 2b:** Posterior-anterior chest X-ray. Postoperative appearence and cervical fusion.

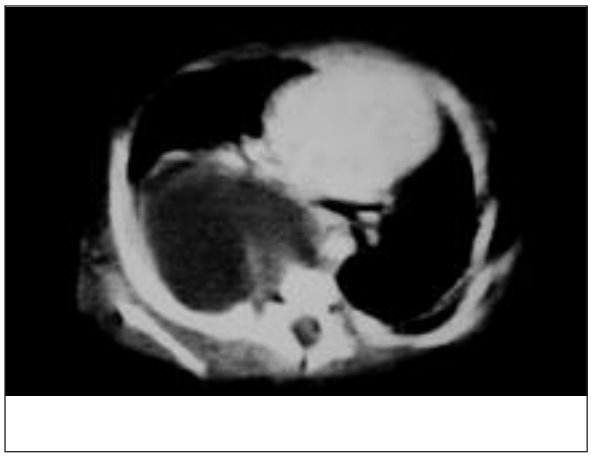

**FIGURE 3:** Thorasic CT: a round cystic lesion of 6x6 cm with smooth surfaces is seen at the right lung.

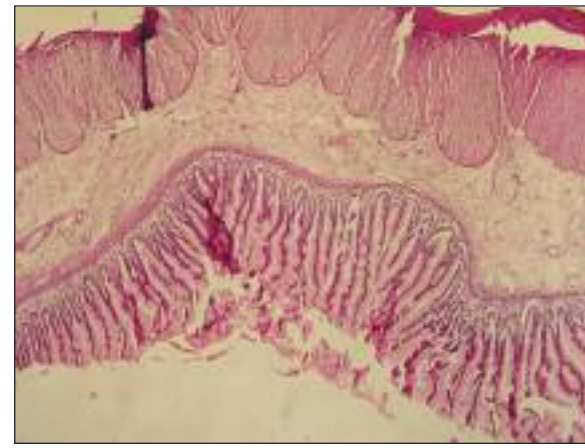

**FIGURE 4:** Pathological examination of the cystic material. A cystic formation with fibromuscular wall lined with intestinal epithelium having villous protrudings towards the surface (20x20, x100 times, HE staining).

tomatic. Vertebral anomalies associated with posterior mediastinal cyst is a known feature of enteric cysts.[1] The patient was admitted to our clinic for respiratory distress and with a lesion replacing two thirds of the right lung and fusion on the thoracal vertebrae, which was observed on posteroanterior chest X-ray.

Klippel-Feil anomaly is known as a triad of symptoms comprising limitation in the neck movements, low hairline, and short neck. This triad is present in at least 50% of the patients.[5] Although uncommon, several pulmonary abnormalities have been reported in association with Klippel-Feil anomaly. Chaurasia and Singh reported ectopic lung in a mature fetus with Klippel-Feil anomaly.[6] Bhagat et al. reported a Klippel-Feil anomaly case with agenesis of the right upper and the middle pulmonary lobes and hypoplasia of the right lower pulmonary lobe.[7] Low hairline and short webbed neck were detected in her physical examination, but Klippel-Feil anomaly was excluded with no detected fusion anomalies in cervical vertebraes.

Spinal anomalies associated with mediastinal cyst are well known anomalies.[8] In 1952, Veenklaas theorised that it separation of the notochord from the entoderm was not complete, a diverticulum of entoderm could be withdrawn from the primitive foregut which would form a cyst and the

attachment of the cyst to the notochord could prevent fusion of the vertebral bodies, resulting in a spinal anomaly.[1] Mortier et al analyzed 26 new patients with multiple vertebral segmentation defects and reviewed 115 previously reported cases. They recognized 3 distinct entities based on radiographic and clinical findings: JLS, a lethal autosomal recessive form, characterized by a symmetric crab-chest; SCD, a benign autosomal dominant condition; and STD, which shows considerable clinical and radiographic overlap with spondylocostal dysostosis and has an autosomal recessive mode of inheritance.[9]

JLS is characterized with spondylocostal anomalies, short webbed neck, low hairline, renal and cardiac anomalies.[10] Respiratory problems caused by intrathorasic meningomyelosel, hernia of diaphragma and thoracic restriction were determined in JLS.[10,11] To our knowledge, mediastinal enteric cyst associated with JLS has been rarerly reported in the literature. In our patient the diagnosis of JLS was excluded with absence renal-cardiac anomalies and, presences of an enteric cyst with no severe symptoms.

Individuals with STD have vertebral anomalies and severely deformed and fused ribs, resulting in the "crablike" appearance of the chest on plain X-ray films. The inheritance pattern is autosomal dominant. Patients afflicted with STD have a higher mortality rate as a result of posterior tethering of the ribs, leading to progressive thoracic restriction as the patient grows.[4] Thoracic restriction was not detected in our patient, but it was thought that it should be observed for developement in the follow-up period.

Individuals with SCD have vertebral anomalies such as hemivertebrae; fused, hypoplastic, and "butterfly" vertebrae; and intrinsic rib defects or malformations (hypoplasia) of varied patterns such as rib fusions and deletions with a non-progressive kyphoscoliosis. Patients with SCD survive more often than those with STD despite gross thoracic abnormalities (possibly because the lungs are not restricted), and it has a reported prevalence of 0.25 in 105 patients.[12] In our patient clinical findings have directed us to a diagnosis of STD in the case.

Mediastinal cysts have been reported be bronchogenic, esophageal, enteric or nonspecific cysts, which originate from the foregut, in origin.[3] Symptoms may change from none to life threatening dyspnea. Standart treatment is surgical resection. Because of the risk of infection and potential for occult malignancy, early resection is advised even in asymptomatic newborns.[13] Our patient had dyspnea and cyanosis. As she had a partial oxygen pressure of 60-80% during monitoring, continuous oxygen was supplied at a rate of 2 L/min. A total cystectomy was performed, and no connection was found between the cyst and the right lung, neighboring organs, or tissues. The lung expanded following surgical treatment and the patient did not require oxygen postoperatively.

This case was reported due to the rarity of the mediastinal enteric cysts, which was associated with costovertebral malformation in the newborn.

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