



Postnatally diagnosed coexisting congenital diaphragmatic hernia with pulmonary sequestration: a report of two cases

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While the associations between pulmonary sequestration (PS) and congenital diaphragmatic hernia (CDH) are known, CDH may be obscured by PS and thus, overlooked on prenatal ultrasonography when coexisting with PS. We present 2 cases of postnatally diagnosed CDH combined with PS. In both cases, PS was prenatally diagnosed as an isolated lung mass, while CDH was confirmed only after birth. Both newborns were sufficiently stable that management was not required immediately after birth. PS may function as an “anatomical barrier” to prevent herniation of the abdominal contents into the chest, thus acting as a “protector” providing normal lung maturation throughout pregnancy. If PS is suspected prenatally, coexisting CDH may be obscured; thus, close prenatal care and counseling of the parents regarding the possibility of CDH are essential. These infants should be delivered at a tertiary center, and imaging should be performed to exclude coexisting CDH.

Keywords: Pulmonary sequestration; Congenital diaphragmatic hernias; Prenatal ultrasonography

Introduction

Pulmonary sequestration (PS) is a nonfunctioning mass of lung parenchyma that lacks communication with the normal tracheobronchial tree. It has an aberrant systemic arterial blood supply, usually from the thoracic or abdominal aorta [1]. Congenital diaphragmatic hernia (CDH) affects 1 in 2,500 newborns and is associated with high rates of morbidity and mortality [2]. While the association of PS with CDH is as high as 30%–40%, the prognosis of their coexistence is unknown [3–5]. We present 2 cases of coexistent PS with CDH. In both cases, only the isolated lung mass was diagnosed prenatally; the coexisting CDH was diagnosed only after birth.

Case report

1. Case 1

A 31-year-old multigravida was referred to our hospital for a fetal lung mass at 27.5 weeks of gestation. On detailed ultrasonography examination, a hyperechoic, homogeneous,

well-defined mass measuring 2.5×1.8 cm was found posterior to the right lower lobe of the fetal lung (Fig. 1A). Because no aberrant systemic arterial blood supply was observed, the initial diagnosis was congenital pulmonary airway malformation (CPAM). The fetal right kidney was slightly elevated, raising suspicion of an ectopic kidney (Fig. 1B). Although the diaphragm deviated upward on the right, it was well

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maintained despite the right ectopic kidney. In the follow-up ultrasonography performed at 34.4 weeks of gestation, persistent left superior vena cava (PLSVC) was noted, but there were no changes in the size of the lung mass and no evidence of CDH.

The female neonate was delivered vaginally at 39.5 weeks of gestation and weighing 2,720 g. Apgar scores were 8 at 1 minute and 9 at 5 minutes. During workup in the neonatal intensive care unit (NICU), her vital signs were stable and oxygen saturation was maintained above 95% in room air. However, the right lung aeration was slightly decreased upon physical examination. Chest radiography demonstrated decreased lung volume and diffuse haziness in both lung fields. One day after birth, right CDH was identified on chest computed tomography (CT) conducted due to the upper deviation of the liver and right kidney seen on an abdominal sonogram. The liver, right kidney, and some bowel loops were located in the right lung field (Fig. 1C). Lung hypoplasia at the right lower lobe and PLSVC were observed. There

was no evidence of CPAM on the postnatal chest sonogram and CT. The CDH was repaired 3 days after birth. A 5-cm Bochdalek's hernia with a sac was present on the posterior side of the right diaphragm. The herniated organs were anatomically reduced into the abdomen. The pediatric surgeon attempted to determine the lung mass based on the prenatal history even though there was no evidence of lung mass on the chest CT. A 5×4-cm lung mass was observed in the upper right diaphragm and the area adjacent to the right of the aorta, and excision of the mass was performed. The origin of the lung mass was unclear, as was its association with other organs. Histopathological examination showed mature and immature lung tissues, which was consistent with extralobar pulmonary sequestration (ELS; Fig. 1D). Two months after the surgery, the patient had no postoperative complications and had appropriate growth for her age.

2. Case 2

A 29-year-old primigravida was referred to our hospital for

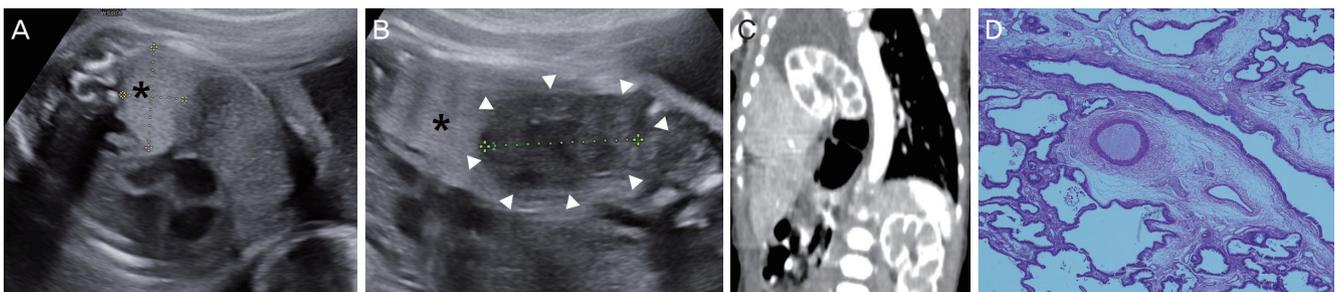


Fig. 1. Prenatal and postnatal findings in Case 1. Prenatal ultrasonographic image showing (A) a hyperechoic mass (asterisk) on the posterior right lower lung on the transverse view and (B) elevated right kidney (arrowheads) beneath the lung mass on the coronal view. (C) Postnatal computed tomography image showing the liver, right kidney, and some bowel loops in the right lung field as well as right lung hypoplasia. (D) Postoperative histopathological findings of the extralobar lung sequestration showing immature pulmonary parenchyma with bronchiole-like structures with pseudocystic dilatation.

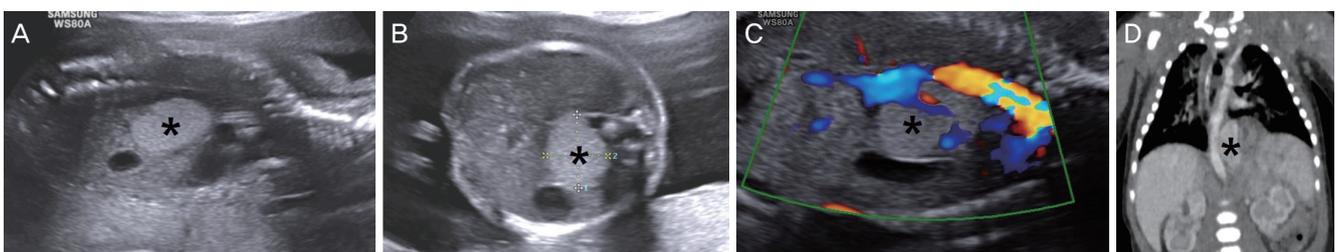


Fig. 2. Prenatal and postnatal findings in Case 2. Prenatal ultrasonographic image showing (A) a hyperechoic mass (asterisk) in the left upper abdominal cavity in the sagittal view and (B) posterior to the stomach in the transverse view. (C) Color Doppler ultrasonographic image showing that the lung mass received blood supply from the descending aorta. (D) Computed tomography image after birth showing the stomach fundus and presumed extralobar lung sequestration positioned below the diaphragm herniated into the left thorax.

a suspected fetal lung mass at 22.2 weeks of gestation. Detailed ultrasonography examination showed a hyperechoic, homogeneous, well-defined mass measuring 2.0×1.8 cm in the left upper abdominal cavity, posterior to the stomach (Fig. 2A and B). The lung mass was thought to be PS, as its blood supply was from the descending aorta (Fig. 2C). Although the stomach was slightly above its normal position, the diaphragm looked intact. On follow-up ultrasonography performed at 35.3 weeks of gestation, the lung mass was unobservable.

The male neonate was delivered through a cesarean section on maternal request at 37.3 weeks of gestation and weighed 2,580 g. Apgar scores were 8 at 1 minute and 9 at 5 minutes. After birth, mild chest retraction was observed; thus, a high-flow nasal cannula (HFNC) was applied. Oxygen saturation was maintained above 95%. Initial chest radiography did not show any abnormal findings. A 2.4×0.5×2.5-cm well-defined, wedge-shaped mass was observed within the left lower thorax on chest CT performed 8 days after birth. The mass was thought to be an ELS because of its arterial supply from the aorta and drainage into the portal vein. A hiatal hernia was suspected because the stomach fundus and presumed ELS (positioned below the diaphragm) herniated into the left thorax (Fig. 2D). Because HFNC was needed for only 2 days after birth and the infant's general condition had improved after ventilatory support, the pediatricians decided to avoid immediate surgery and discharged the infant to the outpatient department. The infant did not undergo the surgery, and no acute illnesses related to the diagnosis was found several months after birth.

Discussion

PS is a discrete mass of nonfunctioning lung parenchyma that does not communicate with the tracheobronchial tree and has an aberrant systemic arterial blood supply, usually from the thoracic or abdominal aorta [1]. Based on the pleural covering, it is classified as intralobar pulmonary sequestration (ILS) or ELS. While ELS is uncommon (15–25% of cases) compared to ILS (75–85% of cases), most prenatally diagnosed cases are ELS, above the diaphragm, and most frequently occur in the lower left side of the chest [6,7]. ELS lesions also occur below the diaphragm and can be confused with neuroblastoma or adrenal hemorrhage [8,9].

ELS is more frequently associated with other anomalies than ILS, including CDH, cardiac abnormalities, pulmonary hypoplasia, and foregut anomalies [1,10]. Among these abnormalities, CDH is the most common coexisting anomaly, occurring in 30–40% of cases [3–5]. CDH is characterized by incomplete diaphragm formation and is complicated by pulmonary hypoplasia and pulmonary hypertension, which may require assisted ventilation or extracorporeal membrane oxygenation (ECMO) in severe cases [11].

The relationship between PS and CDH may be explained by the embryogenetic concept that PS develops at 4–5 weeks of gestation. This occurrence can interfere mechanically with diaphragm fusion and pleuroperitoneal canal closure occurring at 10 weeks of gestation, resulting in CDH [12]. ELS or CDH, when present alone, is often diagnosed prenatally by ultrasonography. However, when both conditions are present, CDH may be missed in prenatal diagnosis. In such cases, an accurate diagnosis may not be possible because the mass effect of PS is speculated to cause an “anatomical barrier” in concomitant CDH, blocking the herniation of the abdominal contents; this may serve as a “protector,” allowing normal development of both lungs throughout the pregnancy [4,13]. CDH may be missed until delivery and then appear dramatically after delivery, depending on the size of the defect in the diaphragm, pressure in the abdominal cavity after labor, delivery mode, respiration of the infant after delivery, resuscitation of the infant, and/or positive pressure ventilation [13]. PS often decreases in size before birth and may not need treatment unless complications such as fetal hydrops or pleural effusion are present after birth [14,15]. The prognosis is generally good if there are no complications; however, the prognosis for cases with coexisting CDH is not established [13].

There were several reasons to explain why CDH was overlooked in our patients. In Case 1, the defect was large enough that the right kidney could herniate into the right lung field. However, the PS was also large enough to prevent herniation of the bowel contents through the diaphragmatic defect. Although the right kidney appeared to be somewhat elevated on prenatal ultrasonography, no evidence of herniation was found. After spontaneous vaginal delivery, CDH was immediately discovered as the anatomical barrier function of the PS was lost; the intra-thoracic pressure was believed to have decreased as the neonate cried, while the increasing intra-abdominal pressure pushed the abdominal contents

into the thoracic cavity. The respiratory function of the infant appeared to be stable considering the degree of herniation. This may be because the protector function of the PS allowed lung maturation. In Case 2, CDH was confirmed later when compared to Case 1 because the infant had no obvious respiratory difficulty and there were no specific findings on initial chest radiography. Chest CT was performed based on the prenatal history of PS; it unexpectedly confirmed accompanying CDH. On prenatal examination, only the stomach appeared to be slightly elevated above the normal position. Considering the PS, we should have suspected the presence of CDH. Because the condition of the infant was stable and he did not show any symptoms of CDH, careful monitoring was performed without immediate surgery.

If a lung mass, especially PS, is suspected on prenatal ultrasonography, coexisting CDH may be obscured. Thus, careful prenatal care and counseling of parents regarding the possibility of postnatal CDH are necessary. Postnatally, imaging should be performed immediately to determine whether surgical treatment is necessary, even if the respiratory function of the infant is stable. Although the natural regression of PS is expected, these patients should be delivered at a tertiary center and coexisting CDH should be excluded.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

Ethical approval

The study was approved by the Institutional Review Board of Kyungpook National University Chilgok Hospital (IRB No. KNUCH 2020-06-026) and performed in accordance with the principles of the Declaration of Helsinki. Written informed consents were obtained.

Patient consent

The patients provided written informed consent for the publication and the use of their images.

References

1. Gerle RD, Jaretzki A 3rd, Ashley CA, Berne AS. Congenital bronchopulmonary-foregut malformation. Pulmonary sequestration communicating with the gastrointestinal tract. *N Engl J Med* 1968;278:1413-9.
2. Langham MR Jr, Kays DW, Ledbetter DJ, Frentzen B, Sanford LL, Richards DS. Congenital diaphragmatic hernia. Epidemiology and outcome. *Clin Perinatol* 1996;23:671-88.
3. Corbett HJ, Humphrey GM. Pulmonary sequestration. *Paediatr Respir Rev* 2004;5:59-68.
4. Grethel EJ, Farrell J, Ball RH, Keller RL, Goldstein RB, Lee H, et al. Does congenital diaphragmatic hernia associated with bronchopulmonary sequestration portend a better prognosis? *Fetal Diagn Ther* 2008;23:250-3.
5. Harris K. Extralobar sequestration with congenital diaphragmatic hernia: a complicated case study. *Neonatal Netw* 2004;23:7-24.
6. Frazier AA, Rosado de Christenson ML, Stocker JT, Templeton PA. Intralobar sequestration: radiologic-pathologic correlation. *Radiographics* 1997;17:725-45.
7. Biyyam DR, Chapman T, Ferguson MR, Deutsch G, Dighe MK. Congenital lung abnormalities: embryologic features, prenatal diagnosis, and postnatal radiologic-pathologic correlation. *Radiographics* 2010;30:1721-38.
8. Kalenahalli KV, Garg N, Goolahally LN, Reddy SP, Iyengar J. Infradiaphragmatic extralobar pulmonary sequestration: masquerading as suprarenal mass. *J Clin Neonatol* 2013;2:146-8.
9. Yang HJ, Lee SW, Lee HJ, Lee JH, Jeon YS. Extralobar pulmonary sequestration mimicking an adrenal tumor. *JSL* 2012;16:671-4.
10. Stocker JT. Sequestrations of the lung. *Semin Diagn Pathol* 1986;3:106-21.
11. Hubbard AM, Adzick NS, Crombleholme TM, Haselgrove JC. Left-sided congenital diaphragmatic hernia: value of prenatal MR imaging in preparation for fetal surgery. *Radiology* 1997;203:636-40.
12. Luet'ic T, Crombleholme TM, Semple JP, D'Alton M. Early prenatal diagnosis of bronchopulmonary sequestration with associated diaphragmatic hernia. *J Ultrasound Med* 1995;14:533-5.
13. Lee MY, Won HS, Shim JY, Lee PR, Lee BS, Kim EA, et al. Protective effect of fetal pulmonary sequestration in two

cases of postnatal manifestation of congenital diaphragmatic hernia. *Ultrasound Obstet Gynecol* 2012;39:719-22.

14. Adzick NS, Harrison MR, Crombleholme TM, Flake AW, Howell LJ. Fetal lung lesions: management and outcome. *Am J Obstet Gynecol* 1998;179:884-9.
15. Cavoretto P, Molina F, Poggi S, Davenport M, Nicolaides KH. Prenatal diagnosis and outcome of echogenic fetal lung lesions. *Ultrasound Obstet Gynecol* 2008;32:769-83.