Prenatal Diagnosis of Bilateral Pulmonary Agenesis: a Case Report

Kyung A Lee, MD
Jeong Yeon Cho, MD
Seung Mi Lee, MD
Jong Kwan Jun, MD
Jieun Kang, MD
Jeong-Wook Seo, MD

We report a case of bilateral pulmonary agenesis (BPA), which was suspected during a prenatal US examination and diagnosed by fetal magnetic resonance imaging (MRI). BPA is an extremely rare congenital anomaly and, although many fetal structural defects can be detected with a high degree of confidence after introducing high-resolution US, the prenatal diagnosis of BPA remains problematic. Other thoracic abnormalities, such as a congenital diaphragmatic hernia, congenital cystic adenomatoid malformation, and pulmonary sequestration, should be excluded from the list of possible diagnoses before coming to the conclusion of BPA, because BPA is absolutely incompatible with extrauterine life, and an accurate internal diagnosis can prevent a futile intervention from being performed.

Bilateral pulmonary agenesis is a rare and lethal condition in which all of those affected die in utero or within the first hour after birth (1-4). Dozens of cases associated with complete agenesis of both lungs had been previously reported (1, 2). Although Vettraino et al. (2) recently authored a report emphasizing the prenatal sonographic appearance characteristics of bilateral pulmonary agenesis (BPA), most of the previous case reports have focused on the macro- and microscopic pathologic conditions (1). Herein, we report a case of BPA diagnosed prenatally with the aid of high-resolution ultrasonography and a fetal MRI.

CASE REPORT

A 28-year-old primigravida was referred to our hospital at 31 + 2 weeks of gestation for further evaluation as a result of abnormal findings on a fetal echocardiography. At 29 weeks of gestation, blindly ended vessels originating from the heart or transposition of the great arteries was first noticed. However, no comments were made regarding any abnormal findings of the fetal lung on a fetal ultrasonography. In addition, the medical and obstetric histories were unremarkable.

An ultrasound examination performed on the day of the first visit at our institute demonstrated an elevated diaphragm, raising the suspicion of associated lung abnormalities (Fig. 1A). The thoracic cavities, especially on the right, were reduced in size. The left thoracic cavity was largely occupied by the heart with a severely left-deviated axis, resulting from the remarkable displacement of the diaphragm cephalad. However, it was not evident whether any lung tissue was present, or whether there were any lung abnormalities or pulmonary veins connecting to the left atrium as well as the right and left branches of the main pulmonary artery. As a result, no specific structural abnormalities of the heart were identified.
Fig. 1. Bilateral pulmonary agenesis.
A. Ultrasound examinations at 31 + 2 weeks of gestation show collapsed thoracic cavity with diaphragmatic eventration. Intact diaphragm (arrow) was noted between elevated liver and narrow thoracic cavity. Right diaphragm was more displaced cephalad than left diaphragm. As result, heart and liver are shown on same plane as axial view.
B. Fetal MRI study on sagittal and coronal views at 35 + 2 weeks of gestation demonstrates decreased volume of thoracic cage, associated with bilateral elevation of diaphragm without any defect. Abruptly ending trachea (arrow) is noted.
C. Postmortem infantogram shows bell-shaped thoracic cage with diaphragmatic eventration.
D. On autopsy findings of thoracic cavity, there are no other structures except for heart, vessels (including aorta), and bilateral masses of fat tissue.
For further evaluation of the lung abnormalities, a fetal MRI study was performed. Similar to the results of the ultrasound examinations, a significantly decreased thoracic volume associated with a bilateral elevation of the diaphragm was identified, thus resulting in an upward migration of the heart and rotation of the cardiac axis. Moreover, no definite lung was identified and the trachea ended blindly near the level of the cricoid cartilage without bronchial branching (Fig. 1B). Together, these findings strongly supported the diagnosis of BPA.

A male fetus weighing 2,530 g was delivered vaginally at 39 + 2 weeks of gestation after the induction of labor and in the presence of neonatologists. The Apgar scores at 1, 5, and 10 minutes after birth were 2, 2, and 2, respectively. Attempts at resuscitation were unsuccessful and after 15 minutes, the cardiac asystole persisted despite all efforts. The neonate died at 44 minutes after delivery. A postmortem infantogram was taken and the results of the study were consistent with the prenatal diagnosis (Fig. 1C). The karyotype of the neonate was 46, XY.

The body size of the fetus was appropriate for its gestational age. There were no congenital abnormalities noted on external inspection; however, multiple abnormalities were noted on an internal examination. The intact diaphragm was elevated bilaterally. A complete agenesis of both lungs was identified along with the absence of lung tissue and bronchial rudiments (Fig. 1D). In addition, neither pulmonary arteries nor veins were identified. An atrial trachea ended blindly at the level of the cricoid cartilage without migration of the heart and rotation of the cardiac axis. Moreover, no definite lung was identified and the trachea ended blindly near the level of the cricoid cartilage without bronchial branching (Fig. 1B). Together, these findings strongly supported the diagnosis of BPA.

In conclusion, an elevated diaphragm and decreased thoracic volume on prenatal ultrasound examinations provide initial clues for BPA. However, it is essential to demonstrate the absence of the right and left branches of the pulmonary artery, as well as the absence of the
pulmonary veins to enter the left atrium by using color Doppler scans. A detailed fetal echocardiography is helpful in detecting major cardiac anomalies and to trace the related vessels. To further differentiate BPA from other congenital abnormalities in the thoracic cage, we recommend fetal MRI as an additional imaging modality for suspected BPA cases from an ultrasound examination.

References
1. Engellenner W, Kaplan C, Van de Vegte GL. Pulmonary agenesis association with nonimmune hydrops. Pediatr Pathol 1989;9:725-730
2. Vettraino IM, Tawil A, Comstock CH. Bilateral pulmonary agenesis: prenatal sonographic appearance simulates diaphragmatic hernia. J Ultrasound Med 2003;22:723-726
3. Nazir Z, Qazi SH, Ahmed N, Atiq M, Biloo AG. Pulmonary agenesis—vascular airway compression and gastroesophageal reflux influence outcome. J Pediatr Surg 2006;41:1165-1169
4. Lee EY, Boiselle PM, Cleveland RH. Multidetector CT evaluation of congenital lung anomalies. Radiology 2008;247:632-648
5. Laudy JA, Wladimiroff JW. The fetal lung. 1: Developmental aspects. Ultrasound Obstet Gynecol 2000;16:284-290
6. Gabarre JA, Galindo Izquierdo A, Rasero Ponferrada M, Orbea Gallardo C, Puente Agueda JM, de la Fuente Pérez P. Isolated unilateral pulmonary agenesis: early prenatal diagnosis and long-term follow-up. J Ultrasound Med 2005;24:865-868
7. Joshi S, Kotecha S. Lung growth and development. Early Hum Dev 2007;83:789-794
8. Moreno-Alvarez O, Hernandez-Andrade E, Oros D, Jani J, Deprest J, Gratacos E. Association between intrapulmonary arterial Doppler parameters and degree of lung growth as measured by lung-to-head ratio in fetuses with congenital diaphragmatic hernia. Ultrasound Obstet Gynecol 2008;31:164-170
9. Kalache KD, Chaoui R, Paris S, Bollmann R. Prenatal diagnosis of right lung agenesis using color Doppler and magnetic resonance imaging. Fetal Diagn Ther 1997;12:360-362
10. Levine D, Barnewolt CE, Mehta TS, Trop I, Estroff J, Wong G. Fetal thoracic abnormalities: MR imaging. Radiology 2003;228:379-388
11. Obenauer S, Maestre LA. Fetal MRI of lung hypoplasia: imaging findings. Clin Imaging 2008;32:48-50
12. Yang JI. Left diaphragmatic eventration diagnosed as congenital diaphragmatic hernia by prenatal sonography. J Clin Ultrasound 2003;31:214-217
13. Jeanty C, Nien JK, Espinoza J, Kusanovic JP, Gonçalves LF, Qureshi F, et al. Pleural and pericardial effusion: a potential ultrasonographic marker for the prenatal differential diagnosis between congenital diaphragmatic eventration and congenital diaphragmatic hernia. Ultrasound Obstet Gynecol 2007;29:378-387